

University of Dundee

## MASTER OF SCIENCE

### Parents' experiences of genetic diagnosis in their child an exploratory study

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Parents' experiences of genetic diagnosis in  
their child: an exploratory study

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Masters Degree  
Dundee University  
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### Declaration

I, Lisa Bryson, declare that this thesis and the work presented in it are my own and has been generated by me as the result of my own original research.

I confirm that:

1. This work was done wholly for a research degree at the University of Dundee
2. Where any part of this thesis has previously been submitted for a degree or any other qualification at this University or any other institution, this has been clearly stated
3. Where I have consulted the published work of others, this is always clearly attributed
4. Where I have quoted from the work of others, the source is always given. With the exception of such quotations, this thesis is entirely my own work
5. I have acknowledged all main sources of help
6. Where the thesis is based on work done by myself jointly with others, I have made clear exactly what was done by others and what I have contributed myself
7. This work has been presented as a poster at the British Society of Human Genetics and the American Society of Human Genetics conferences in 2012.

Signed: .....

Date: .....

## Abstract

There has been a rapid increase in our ability to diagnose genetic conditions in children, using technology such as Next Generation Sequencing and array CGH. Very little is known about the impact of such diagnoses on parents, and the parents' need for support following testing. We, therefore, used a qualitative methodology to explore the effect of diagnosis on parents and to identify areas in which additional support would be most useful.

Semi structured interviews were undertaken with 11 parents of children with a confirmed genetic diagnosis causing developmental delay and additional syndromic features. All patients were identified and recruited with ethical approval and consent from within NHS Tayside. The interviews were transcribed and analysed using framework analysis.

Four main themes were identified: the reaction to having a genetic diagnosis; how they felt the healthcare team managed them; the support that was available and how a genetic diagnosis affects the family. Within each theme, there were multiple sub-themes highlighting both positive and negative effects on the family. A key point that emerged was that parents felt there was a lack of signposting to support especially before and immediately after diagnosis, and that provision of appropriate information and structured follow up would improve parents' experiences.

Our results have allowed us to design a questionnaire to ascertain how often key themes are appropriately dealt with during consultations, and to propose potential management changes required to improve follow up of parents with a child with a new genetic diagnosis.

## Background

### Diagnosis of Genetic Disorders

Developmental delay occurs in 5-10% of the childhood population (1) and has a profound impact on both the person affected and their family. There are many different causes, including infection, metabolic abnormalities, and anoxic injury at birth. Genetic mutations are a common cause of learning disability. Some of the most common genetic conditions can be investigated easily, for example karyotyping for Down's syndrome.

Genetic diagnosis is becoming much more efficient (2). In the past 20 years genetic tests have become more accessible and commonly used. Previously diagnosis was based on the study of dysmorphology (3). Cytogenetic and molecular techniques are constantly improving. Since the completion of the human genome project, it has been suggested that more genetic testing will gradually become available (2, 4, 5). Additionally, it is becoming cheaper and faster to sequence a human genome as a result of the developments in DNA sequencing technologies. It is likely that these developments will lead to changes in clinical practice and the way in which genetic diagnoses are found (6).

It has been estimated that 50% of children who have a severe learning disability (7) and 60% of children who have multiple congenital anomalies do not have a definite diagnosis (8). In the future this may not be the case due to the advances in the field of genetics. The age at diagnosis is falling significantly in certain syndromes, for example Prader Willi and Fragile X (9, 10). Earlier diagnosis means that appropriate multidisciplinary team members can be involved in the case and a management plan can be made earlier rather than later; such as the use of growth hormone treatment for children with Prader Willi syndrome (9).

Newborn screening is continually developing. In the 1960s, Phenylketonuria (PKU) was the first genetic condition to be tested for as a newborn (11). Currently there are a number of genetic conditions which are screened for at birth, such as PKU, and many more are becoming available (12). The increasing availability of genetic investigations will lead to earlier diagnoses for many families. It has been suggested that having a well organised screening programme for genetic conditions at birth, such as Fragile X, would allow



information, counselling and treatments to be provided in a more successful and time critical manner before, during and after diagnosis (10).

The Deciphering Developmental Delay (DDD) study has been using microarray, whole genome sequencing and DNA microchip technologies to discover the genetic mutations involved in causing developmental delay. Children with conditions such as autism may have genetic mutations, which have not yet been discovered, whilst some children have been diagnosed clinically with syndromes in which the mutation has never been found and other children do not have diagnoses at all. Although genetic conditions are common in total, each condition is rare. Genetic data collected during the DDD study is being analysed and compared using the DECIPHER database, which is an international database that helps link genetic mutations to medical conditions. This will help to find diagnoses for children with developmental delay and gain a better understanding of the genetics behind it (13).

Therefore, with our growing knowledge of genetics and genetic conditions and increasing ease of diagnosis, it is going to become more important to develop optimal management pathways for the children with genetic conditions.

Parents' expectations of diagnosis have been identified in previous research. The DDD study had an online discussion forum, interviews and focus groups (13). From these, they found that the participants would welcome a label or an 'answer'. Many parents suggested that an end to the search would help them to accept their child's condition. Having a label would allow parents to explain the condition to family and friends, providing them with answers on risk of recurrence (13). Lewis et al interviewed parents of 9 children without a diagnosis and found that parents wanted confirmation that there was something wrong with their child in order to legitimise their concerns (14). Lenhard et al collected questionnaires from 579 mothers of children with Down's syndrome, mental retardation of unknown origin and without disability. They found that some parents felt something must have gone wrong in pregnancy or at birth (15). This caused feelings of guilt. Additionally, the diagnosis has been found to allow better understanding by the healthcare team and can alter management (13). A diagnosis may help to gain an understanding of what is to come in the future (16).

As well as providing the family with a cause and potential management options, research suggests that having the diagnosis may make it easier to find help. It has been suggested

that the diagnosis allows family members to speak to other families affected and access specific support groups and services available. Lenhard et al. found more parents of children with Down's syndrome accessed support groups than parents of children who did not have a known diagnosis (15). Mothers of children who did not have a diagnosis were less likely to realise that many other parents were in a similar position (15).

Despite these benefits, drawbacks in gaining a diagnosis have been found. In interviews with parents of 43 children affected by a diagnosis of Ataxia Telangiectasia, it was found that having a label and knowing more about the disease could lead to loss of hope. Parents were no longer able to 'pretend that everything is fine' or that their child would grow out of the condition. Being given a diagnosis of Ataxia Telangiectasia was described as being similar to getting a 'death sentence' (17). In addition to this, having a diagnosis may only be helpful if it means something to the parents. One parent, who took part in an interview with Lewis et al, was told that her child had a 'translocation'. She felt that knowing this did not help her. She compared it to being told something in a foreign language (14).

There can be a great deal of variation within conditions themselves. This brought hope to one parent reflecting on her own experiences of genetic diagnosis in her sons affected by Adrenoleukodystrophy. She described her belief that "hope is uplifting and valuable" (18). However, this variation lead to increased anxieties in some parents of children with Neurofibromatosis 1 as it added to the unknown of what the future will hold (19).

It has been found, from interviews with parents who do not have a diagnosis for their child, that receiving a diagnosis may not be a priority as it would not change their child or the management (14). In interviews with 19 parents of children who did not have a genetic diagnosis for hearing loss, they similarly felt that it was more important to take care of their child than find the gene that caused the condition (20). This feeling was more common when there was a family history of the disability. If there was no family history then there appeared to be an increased interest in finding a genetic diagnosis.

How parents cope without having a diagnosis for their child with developmental delay has been researched (14, 21). Feelings of grief were found to be similar to those who have a diagnosis. However, without a diagnosis there were additional feelings relating to the unknown. Parents of 9 children with different syndromic features without a diagnosis

reported anxiety in relation to the child's prognosis and recurrence of a condition (14). Additionally, parents of 16 children with unidentified multiple congenital anomalies suggested in interviews that it may be more difficult to come to terms with the condition without a diagnosis and they may feel less in control (21). Some parents felt that acceptance of the child's condition may take longer without a diagnosis (21). One parent of a child with syndromic features but with no diagnosis mentioned the added practical difficulties when a diagnosis is not known. For example, housing and education may be difficult to organise if the course of the condition and prognosis cannot be described (14).

It has been found, from parents' and children's reports on chronic illness, that the time between parents noticing something is wrong with their child and receiving a diagnosis is particularly stressful for families (22). Ataxia Telangiectasia can be wrongly diagnosed as Cerebral Palsy and it has been reported to take years to get a real diagnosis. These delays in diagnosis have been found to have a negative impact on the families involved (17). In one parent's reflection upon her experiences of getting a genetic diagnosis of Adrenoleukodystrophy in her children, she discussed how difficult it was to wait for test results and then wonder whether the siblings were similarly affected (18). This demonstrates the difficulties associated with the unknown. Additionally, parents in a previous qualitative study explained that the worst possible outcome of a genetics referral would be not to obtain a definite answer (16). Thus receiving a diagnosis with a bad prognosis can be perceived as better than not receiving one at all.

The importance of receiving a diagnosis for a child has been highlighted both when paediatricians were asked about improving management and when parents were asked about receiving a diagnosis of Down's syndrome after birth (23-24). Health professionals emphasized the importance of an appropriate setting and time when providing a diagnosis to allow for optimal communication. Additionally, having a good understanding of the family's background was recommended (23). Both papers on this topic document the benefit of providing appropriate information and follow up. Parent satisfaction has been found to directly relate to the empathy and communication provided at diagnosis of developmental disability from interview analysis by Hasnat and Graves (25).

Results of a postal questionnaire study, directed to parents of children with Fragile X syndrome, found that parents were given diagnoses in many different ways with varying success (10). Some parents felt that they were not given any information on what the diagnosis meant whereas others had very supportive and empathetic experiences. Another study found that follow up was not given routinely to children diagnosed with Neurofibromatosis 1 although it was felt to be needed by parents (19). Follow-up was thought to be very important as it allowed the parents time to formulate questions and gave them an opportunity to discuss anything that they did not understand.

Parents are sensitive at the time of diagnosis. One paper described how a parent felt when asked if photographs could be taken of her son for clinical reasons (16). She felt unable to say no to the request and felt that they were not treated as well as she wished. In another paper, a parent described feeling that some language, which was used by the health care professional, made her feel like her sons were “freaks” (20).

Emotional themes involved with genetic diagnosis have been identified in previous qualitative research. Anxiety, worry about children, guilt, anger, uncertainty, sadness and grief, depression, and redemptive adjustment were discussed repeatedly in 7 focus groups and 19 interviews with patients with a genetic diagnosis or patient representatives (26). These results may be intuitive for the large part. However, this research highlighted how parents may feel when getting a genetic diagnosis and the emotional repercussions it has.

The mourning process has been discussed in a number of Studies. One study, which explored the experiences of 18 parents of children with Neurofibromatosis 1, described the “shock, fear, and subsequent depression” that could be felt (19). Interviews on newborn screening for Cystic Fibrosis suggested that this time was very emotional for parents and it could cause a reaction similar to post-traumatic stress (27).

Furthermore, from Leventhal’s research with parents of children without a diagnosis, in the longer term lack of sleep, money worries and constant hospital appointments have been reported to be major contributing factors to parental stress levels (14). Adaptations need to be made to the parents’ normal lives, including work, children’s schooling and the family’s housing, in order to ensure their child’s needs are met. Additionally, parents reported feeling stigmatised within society and changing friendships as a result of the diagnosis. All of

these factors appeared to add to the emotional effects of having a child with a genetic diagnosis (14).

Another topic, which may appear to be relatively intuitive, is parents stress and fears related to the future. Research, which looked into how parents who have a child with Ataxia-Telangiectasia feel about the diagnosis, discussed parents imagining their children growing up and becoming more independent. However, for these parents, who have a child with a genetic condition causing a severe disability, the children may grow and become more dependent on their parents. One set of parents noted how unusual it was to feel terrified that their child would outlive them (17).

It has also been suggested that there may be differences in the way fathers and mothers deal with their emotions. One study, which used a qualitative approach to determine the psychological impact of genetic testing on parents, described finding mothers to be more anxious in relation to their child's condition compared to fathers. They hypothesized that this may relate to mothers acting more commonly as the main carer, attending the clinic appointments and relaying the information to other family members (28). In further research, fathers were thought to be less emotional, more distant and more likely to be in denial than mothers when interviewed about dealing with a child's diagnosis of Neurofibromatosis or Ataxia Telangiectasia (17, 19).

Many reasons have been found to explain why diagnosis is difficult for families. Parents of children with Ataxia-Telangiectasia described mourning for the child they had before the diagnosis, the child that they thought they had and the child that they were never going to have (17). These parents described different coping mechanisms, which they employed in order to come to terms with the diagnosis. Some examples of this include turning their feelings of grief into feeling special, as they were lucky enough to have a child who was different (17). Others benefited from helping people who were in the same situation as themselves, for example becoming involved in fundraising (17). It was noted that families who held strong religious beliefs and had a child with Neurofibromatosis 1 appeared to have a more positive outlook on the diagnosis (19).

Furthermore, research, looking at how parents cope without having a genetic diagnosis for their child, has identified alternative coping methods. Some parents tried to remain positive

and compare themselves to people who were worse off (14). Some found that being in touch with families who were in similar situations was beneficial (14). In another paper, some parents of children with multiple congenital anomalies found it helpful to try and find out as much as possible and get involved in as many groups as they could find (21).

Coyne and Canam discussed the coping mechanisms used by parents of children with chronic conditions including Cystic Fibrosis and explained the potential benefits of denial (29, 30). They described denial as being a short term strategy while parents developed longer term coping mechanisms (29). If denial remained, it could affect the parents' ability to deal with a situation effectively.

Hoare et al. considered the reasons why parents have different reactions and coping mechanisms. They achieved this by using a survey and analysing the psychosocial adjustment of parents of children with severe intellectual disabilities. They found that parents who had a higher education level and were home owners were more susceptible to stress (31). Therefore, it was hypothesized that parents of a higher socio-economic background may not adapt and cope as well when compared to parents from a lower socio-economic background (31). They also found parents who became stressed were more likely to access respite care. Accordingly, parents who did not access respite care were found to have a more positive outlook on their coping abilities (31).

Questionnaires, completed by parents of children with a range of different conditions causing developmental delay, confirmed that the condition that the child suffers from affects how parents cope (31-33). Hoare et al. found that a carer's mental health can be negatively affected by increasing severity of the child's condition (31). Another study, which explored how parents of children with Down's syndrome, Fragile X syndrome and Autism cope, found that the biggest predictor of maternal outcomes was the behavioural symptoms that the child expressed (32).

Parents of children with certain rare genetic conditions have higher levels of anxiety and depression, as found when analysing questionnaire results from parents of children with Angelman, Cornelia de Lange and Cri du Chat syndromes (33). It has been suggested that the rarity of the condition may add to parents stress levels (33). It has been found, from a review of parents' and children's reports of illness, that feelings of uncertainty in relation to

illness are associated with increased anxiety (22). It could be hypothesized that the rarity of the genetic condition may cause increased anxieties because there are more uncertainties. Down's syndrome is a relatively well-known diagnosis with good access to support. Lenhard et al. found parents of children with Down's syndrome were as emotionally well as parents with children who did not have any disabilities (15).

### Medical Professionals

Genetics is a specialised area of medicine. It has been reported in Skirton's research that parents can feel that the healthcare team who are managing them and are giving them advice on genetics may lack knowledge in the area (16). Many studies have reported that paediatricians may have difficulty supplying information on conditions that they do not know a great deal about and information may be better supplied by specialists in the area who are trained in genetic counselling (34, 35). Only 7.4% of 230 parents interviewed with a child with a genetic metabolic disorder reported seeing a genetic counsellor, hence other health care professionals delivered genetic information (36). Qualitative research on rare genetic syndromes has shown that a lack of knowledge from the healthcare professional leaves parents feeling unsupported and can lead to important investigations being missed (37, 38)

In a study with 18 parents of children with a diagnosis of Neurofibromatosis 1, it was found that a quarter of the families were referred to a genetics department (19). The families who were not referred and were diagnosed by alternative physicians appeared to report a more negative experience at diagnosis. Within the genetics department, the trained health professionals knew how to manage families optimally and it was found that genetics specialists were able to promote a more positive outlook on the prognosis of the condition.

In addition to this, neurologists, who may not be trained in genetic counselling, have also been found to leave a more negative memory of the genetic diagnosis than qualified genetic counsellors. Results from a questionnaire developed for parents of children with Spinal Muscular Atrophy found that neurologists were able to provide a great deal of factual information. However, they were thought to have the poorest understanding of parents'

needs when providing genetic counselling (39). On the other hand, from a questionnaire to parents of children with Fragile X syndrome, it was found that parents who do receive genetic counselling often still have difficulty retaining information gained during consultations (10).

Skirton found that families are often not given enough information on what to expect from genetics services when they are referred (16). Some parents have reported fear and anxiety when they heard the word genetic as they felt they were to blame for the child's condition. These problems were thought to be due to a lack of knowledge from individuals who were making the referrals and not providing appropriate information (16). Additionally, Barr and Millar discovered that failing to provide this information can leave parents feeling they have not got the most out of their genetics appointment (40).

There has been a great deal of research on genetic counselling consultations, to ascertain optimal practice. It is thought that by allowing the patient to lead the consultation, an environment in which the parent can let concerns flow easily is developed and this allows psychosocial worries to be investigated fully (41, 42). Additionally, technical jargon was found to be intimidating and suppressed the 'emotional voice' of the parent. It is believed that patients remember more of what they have been told if they gain an understanding and are given the opportunity to ask questions. Additionally, if not given this opportunity then their anxiety levels increase and they were less likely to comply with their management plan.

### Access to support and information

A number of issues have been highlighted in previous research with regards to support provided by the healthcare team to families affected by a genetic diagnosis. For example, Lewis et al analysed parents' recollections of healthcare team management. Consultations with many different healthcare professionals were described, as the team involved was often extensive. Parents' main problem with this appeared to be the need to repeat their child's story. This was reported as being distressing for some of the parents (14). In contrast to this, some parents in a different research study by Skirton described the benefit of



repeating their story. It gave them a chance to relieve some of their emotions and talk to someone who wants to listen and would understand the condition and their situation better than their friends (16).

In addition to this, parents explained that having a large team involved in the child's care lead to difficulties in knowing whom to turn to for support (14, 16). Some parents suggested that having a key worker, who could help to ensure the needs of the family were being met, would be of significant benefit. Furthermore, 24 paediatricians were asked to discuss management of children with severe disability and they appeared to agree that there was a need for a key worker to be involved (23). Lewis et al concluded that a key worker could be in charge of coordinating care, forming a care plan, supporting the family, reviewing their needs and speaking to services and schools on their behalf (14).

Qualitative research, in the form of interviews, questionnaires and focus groups, has examined the benefits of parent led support groups for children with disabilities. From this research, it was identified that there are many ways these groups can provide support. They were described as providing parents with new skills and a sense of power, confidence and belonging (43, 44). Parents reported gaining control of their world and feeling understood within a community. This was achieved by parents helping each other through everyday life in a way that could not be achieved by those who did not have a child with additional needs. Having this social network allowed them to feel "normal". The support groups also aided acceptance. Parents emphasized the importance of being able to speak to someone who had been through a similar experience. In spite of these benefits, parents involved in Skirton's research on being referred to genetics, highlighted a lack of referrals or signposting to this type of support by the healthcare team (16).

Furthermore, parents, who were given a diagnosis of a sex chromosomal condition in pregnancy, reported the importance of meeting other families affected by the same condition as it provided similar benefits to those mentioned previously. Additionally, couples appreciated meeting parents of children with similar conditions as it helped them grow a better understanding of what to expect in the future (34). In contrast to this, parents of children with Ataxia Telangiectasia differed in how beneficial they found this (17). For some parents it increased their anxieties and fears when they saw the future for their child.

A number of papers discuss the information that is provided to parents when they are receiving a genetic diagnosis or undergoing genetic testing. Many issues have been highlighted with regards to the amount, reliability and relevance of the information and the understanding and support that parents gain. For example, in one study parents of children with sex chromosome abnormalities were asked about their understanding of the child's predicted syndrome after first receiving information on the diagnosis. It was found that 77% (twenty) of parents considered themselves to have a "poor" understanding (45).

One study looked into the written information that was provided to patients undergoing genetic testing. They identified what information is usually included and what is often forgotten (46). The condition being tested for, how it can be managed and hereditary details were usually included. However, only 50% of the information sheets involved in the study provided details on where to get extra information or support. Less than half of the information sheets provided information on what happens after testing, what the patient's rights are and the possible psychological risks involved with genetic testing. It was also found that the benefits of testing are much more likely to be discussed than the risks.

This study also compared different ways in which information can be provided (46). From this they identified that leaflets were a lot more informative than letters written from the genetics clinic. However, in another study by Barr and Miller, parents who were sent letters documenting what was discussed within a consultation were found to have a better understanding of a diagnosis (40). Also, research, based on the information that is provided to pregnant women with suspected foetal anomalies, found that anxiety levels decreased when information was provided in the form of a written letter after a consultation. The letter may have provided a resource to go back to at a later date (47).

Overloading parents with information at the time of diagnosis has been reported to cause further distress at an emotional time. Collins et al found, from 14 interviews with parents of children with Cystic Fibrosis and Down's syndrome, that the information given to parents receiving a genetic diagnosis in their child may not be suitable (48). They hypothesized that information overload, which caused a lack of understanding, lead to this result (48). A qualitative study on genetic testing for children with hearing loss identified a need for healthcare professionals to listen to what the parents want to know (20). Similarly, in one of

the interviews in Hasnat and Grave's research, a parent discussed the importance of being allowed to judge the level of information that they could handle and decide what they wanted to know about their child with developmental delay (25).

Interviews, which were conducted on adolescents and parents about genetic testing, found that the general public have a poor understanding of genetic testing and the Internet would be the first place they would look to find information (49). The main drawbacks in searching for information on the Internet include the inconsistencies in the facts, difficulties finding a reliable and understandable source and harm done in overconsumption of information (50). Previous research has found that if patients understand a sufficient amount, then they may not need to look up resources in the media or the Internet, which can be confusing, inaccurate or exaggerated (51). This research has led to the importance of communication skills being stressed in training programs for medical staff.

Communicating the genetics of a condition and the risk of recurrence has been found to be important to parents as they do not always gain a good understanding. Parents in Fanos and Mackintosh's study were found to underestimate the risk of having further children affected by Ataxia Telangiectasia significantly (17). Additionally, parents of children with Cystic Fibrosis have been found to lack a firm understanding of what being a carrier means (27, 52). One study reported that 57% of parents, of children who have been diagnosed with Cystic Fibrosis, understand that their child has a 50% chance of having a child with Cystic Fibrosis, if they reproduced with another carrier (52).

One mother's reflection on her sons' diagnoses with Adrenoleukodystrophy discussed how her extended family was also affected by the diagnoses. She had the role of being the main information provider. Thus, when she received information from the genetics department, she needed to relay it to her family members (18). She highlighted how difficult it was for her to come to terms with the diagnosis whilst trying to answer her family's questions, as they were also concerned. She suggested being given written information for family members would have been helpful (18).

### The wider family

There are many ways in which genetic conditions can affect families. A number of research articles touch on these. For example, family members have been found to act as a supportive network for parents affected by a genetic diagnosis in a child. Questionnaires completed by parents of children with rare trisomy conditions identified that the partners of mothers act as a particularly important support (53). However, family experiences differ widely and the result of having a genetic condition within the family can cause friction.

Bostrom and Ahlstrom identified, from interviews with individuals affected by Muscular Dystrophy and their next of kin, that the hereditary aspect of Muscular Dystrophy was an important concern for the whole family that lead to feelings of guilt and blame (54). In another paper, on the consequences of receiving a false positive result in new born screening for Cystic Fibrosis, it was highlighted that the identification of a gene mutation lead to blame being placed. It also caused strain on the relationships, which could affect the whole family (27).

Similarly, McAllister et al identified feelings of guilt and blame from focus groups and interviews involving 52 individuals involved with the genetic services (26). They found that some patients opted not to disclose the genetic diagnosis with their family, as they felt it might be an easier option. This reportedly caused further feelings of anger to develop within families when the diagnosis was discovered (26).

Apart from blame and guilt causing friction, the added stress of having a child with additional needs appears to put added strain on family relationships. Wei and Yu identified the effects of having a child with disabilities on parental relationships and employment by using a parent and school staff survey of 9000 students in Special Education (55). This survey reported that the families' experiences varied significantly depending on the type of disability. Single parent families were more common if a child had 'mental retardation, emotional disturbances, or multiple disabilities'. Additionally, mothers were more likely to be unemployed if they had a child who suffered from 'hearing, visual, and orthopedic impairments, autism, traumatic brain injury, and multiple disabilities' (55).

Lewis et al, who researched families' experiences without a diagnosis for their child with additional needs, found that friction could develop between siblings as well. This is thought to relate to the effect that having a sibling with additional needs can have on childhood (14). Reportedly, it can restrict family outings and, as a result, some siblings were not able to have the same experiences as their peers. Parents felt that this may have caused resentment.

Having a child diagnosed with a genetic condition affects parents' decisions about future reproduction. A number of studies have examined the choices that parents make and have tried to understand the reasoning behind them.

Read reported that stress levels, in 230 parents of children with genetic metabolic disorders, were directly related to how parents felt about having future children (36). It affected their opinion on whether or not they would consider prenatal genetic diagnosis or termination and whether they would want to avoid having children with the same condition in the future (36). Parents were more likely to consider prenatal diagnosis (56%) than termination of an affected pregnancy (10%). Prenatal diagnosis was thought to help parents prepare for what was coming or help them make a decision on termination. It was also found that parents who wanted to prevent having future children with the same condition and would consider prenatal testing and termination had a higher parenting stress index, children with a lower score on an adaptive behaviour scale, less social support and more concerns for their child's future (36).

Furthermore, a retrospective study looking at data records from 61 couples, who were told their child would have a sex chromosome abnormality, found that the karyotype of the child, the age of the couple and the number of previous children were major influencing factors when it came to deciding whether to continue or terminate a pregnancy (56). The couple were more likely to continue the pregnancy if they were older and if they had no other children.

In a further study from Exeter University, 43 parents were interviewed about their reproductive choices after having a child with a genetic condition, with respect to prenatal testing and termination. It was thought that the vast majority of parents in the study avoided facing the choice of whether to terminate. They did this by either deciding not to

have further children or not to have testing during pregnancy. Many reasons behind these decisions were given, ranging from the risks involved in testing, to feeling that medical professionals do not know everything and would not provide them with the answers that they would want (57).

In a reflective article by McGowan, she discussed her experiences of being a mother of a child with a genetic diagnosis. She described her disappointment at not having a genetic counsellor to speak to about the ethical issues associated with some of the management options available relating to future reproduction, for example amniocentesis and termination. She felt she did not have anyone to speak to about the emotional repercussions on this subject, although she was given the factual information that she required (18). Questionnaire results from parents of 73 children with Spinal Muscular Atrophy found that many parents decided not to have more children as they assumed that there was a large risk. These assumptions were often made without seeking any facts. This was thought to be a result of not having access to genetic counselling (39).

Advances in the field of genetics have resulted in an increasing ability to provide families with a genetic cause for their child who is developmentally delayed. This has led to parents receiving rare genetic diagnoses. Previous research has found that parents can have difficulty coping with a genetic diagnosis and often lack understanding of what it means for them. Additionally, the healthcare team may not manage these families in the most appropriate way. Therefore, how parents feel they are managed within NHS Tayside is of interest to healthcare professionals who are involved with these families. This highlighted the need for further research to be carried out into parent's experiences in receiving a rare diagnosis and how they should be managed in order to improve these.

### Aims of current study and Research Question

Little is known about parents' reactions to a rare genetic diagnosis in their child and how the parents' support needs are managed by the healthcare team during follow up. Previous research, exploring parents' reactions to a genetic diagnosis, has identified some support needs and shown that management currently given to these families is often suboptimal. This research aimed to explore parents' experiences of genetic diagnosis within one regional NHS health board. Specifically, what parents needs were around the time of diagnosis, how they felt they were managed and potential areas for improvement.

Research question: How do parents within Tayside perceive they are managed by the healthcare team and what changes could benefit them?

## Methods

### Literature Review

A systematic approach was taken when searching for journal articles within the literature. Medline was used as the main database. The terms that were used were genetic testing, diagnosis and counselling, congenital abnormality, developmental delay, parent psychology, child and support. Table 1 includes the articles that were of most relevance to this study based on their abstract. Research that examined the management and experiences of families of children with genetic conditions were included. Additionally, studies on children with developmental delay were included if the abstract was thought to be relevant. Research was excluded if the genetic conditions did not affect the child until adulthood or related to cancer genetics. This left 23 journal articles which were included as part of the systematic review of the literature. From these articles further sources were identified from the references.

Table 1. Peer reviewed papers included in the literature review.

Title	Authors	Method	Participants	Year
Living Without a Diagnosis: The Parental Experience	Lewis C et al.	Interview	Parents of 9 children with different syndromic features without a diagnosis	2010
Psychological Benefit of Diagnostic Certainty for Mothers of Children With Disabilities: Lessons From Down's Syndrome	W. Lenhard et al.	Questionnaire	579 Mothers of children with Down's syndrome, with a mental retardation of unknown origin or without disability	2005
Parental experience of a paediatric genetic referral	Skirton H	3 interviews each	Parents of 20 children with different suspected or confirmed genetic diagnoses	2006



Never again joy without sorrow: The effect on parents of a child with ataxia-telangiectasia.	Fanos JH, Mackintosh M-A.	Questionnaire and Interview	Parents from 43 families affected by ataxia telangiectasia	1999
Beyond the disorder: one parent's reflection on genetic counselling.	McGowan R.	Reflection	One parent with 2 sons with adrenoleukodystrophy	1999
Parents' responses to their child's diagnosis of neurofibromatosis 1	Ablon J	Interview	Parents of 18 children affected by Neurofibromatosis 1	2000
Parental narratives on genetic testing for children with hearing loss: A qualitative inquiry	Steinberg AG et al.	Interview	Parents of 19 children undergoing genetic testing for hearing loss	2007
Parental attitudes toward a diagnosis in children with unidentified multiple congenital anomaly syndromes.	Rosenthal ET et al.	Interview	Parents of 16 children with unidentified multiple congenital anomalies	2001
Disclosure of developmental disability: A study of parent satisfaction and the determinants of satisfaction	Hasnat MJ, Graves P	Interview	Parents of 23 children with developmental disability	2000
Going a different road: first support and information needs of families with a baby with Down's syndrome	Muggli EE et al.	Interview	Parents of 18 children affected by Down's Syndrome	2009
The psychological impact of genetic testing on parents	Dinc L, Terzioglu F	Interview	Parents of 128 children having genetic testing for development delay	2006

Reproductive decisions of parents of children with metabolic disorders	Read CY	Interview	Parents of 230 children with genetic metabolic disorders	2002
Spinal Muscular Atrophy Genetic Counselling Access and Genetic Knowledge: Parents' Perspectives	Meldrum C et al.	Questionnaire	Parents of 73 children with Spinal Muscular Atrophy	2007
You Have to Sit and Explain it All, and Explain Yourself.” Mothers’ Experiences of Support Services for Their Offspring with a Rare Genetic Intellectual Disability Syndrome.	Griffith G et al.	Interview	Mothers of 8 adult offspring with diagnoses of Angelman, Cornelia de Lange, or Cri du Chat syndrome	2011
A sociolinguistic exploration of genetic counselling discourse involving a child with a new genetic diagnosis	Babul-Hirji R et al.	Recorded genetics counselling	10 genetic counselling sessions with genetics counsellors and parents of children with genetic diagnoses	2010
Living with a hereditary disease: persons with muscular dystrophy and their next of kin.	Bostrom K, Ahlstrom G	Interview	46 people affected by muscular dystrophy and 36 next of kin	2005
A community survey of children with severe intellectual disability and their families	Hoare P et al.	Interview and Questionnaire	Parent or carer of 145 children with severe intellectual disability	1998
Psychological Well-Being and Coping in Mothers of Youths With Autism, Down’s or Fragile X Syndrome	Abbeduto L et al.	Questionnaire measures of depression, behaviour...	Mothers of children with Autism, Down’s Syndrome, or Fragile X Syndrome	2004

Psychological well being in parents of children with Angelman, Cornelia de Lange and Cri du Chat syndromes.	Griffith GM et al.	Questionnaires	Parents of 69 children with different rare conditions and Autism	2011
The perceived effects of parent-led support groups for parents of children with disabilities.	Law M et al.	Interviews	20 parents with children with disabilities involved in 9 different parent led support groups	2001
The Benefits of Mutual Support Groups for Parents of Children With Disabilities	Solomon M et al.	Questionnaire and focus group	56 parents from 6 groups (questionnaire) and 43 parents from 5 groups (focus group)	2001
We've been through it all together': supports for parents with children with rare trisomy conditions.	Bruns D, Foerster K	Qualitative survey	20 parents of children with rare trisomy	2011

### Interviews

A basic framework for the interviews was developed after deciding the key areas that were to be covered with the parents. Semi structured interviews were used as they provide use of a framework and also freedom to adapt the interview depending on the context. This technique allowed further insight into the parents' experiences. The parents were given a two-way conversation and were able to build a rapport with the interviewer while discussing sensitive topics.

Eleven mothers and one father were interviewed for this study. They were all parents of a child diagnosed with a rare genetic condition causing developmental delay and additional syndromic health problems. The aim was to discover how parents reacted to their child

being diagnosed with a rare genetic condition and to identify key points in the parents' experiences which should be addressed when giving parents follow-up advice for the future and managing their cases in general. Further details of the parents involved are provided in table 2.

### Ethics approval

The study was conducted in accordance with approvals from NHS Tayside Research and Development (R&D) and Research Ethics Committee (REC) from the 13<sup>th</sup> of January 2011. The REC reference number was 10/S1401/64 and the Tayside R&D reference number was 2010GE08. Under the requirements of the Scottish Executive Health Department's Research Governance Framework for Health and Community Care, the University of Dundee agreed in principle to act as sponsor for this project. The application number was 00000061 and was approved from the 10<sup>th</sup> of October 2010. All documents are attached in the appendix.

Two amendments were made during the course of the study. The first was to increase the number of participants, the duration of the study and to include patients who were given a diagnosis within a year from interview. These participants were excluded previously. The second amendment was the introduction of the structured questionnaire for use with the consultant paediatricians and geneticists within Tayside. The results from this are included in the appendix.

### Participant selection

The participants selected to take part in this research study had children who had been diagnosed with a genetic condition. The genetic conditions were rare and caused developmental delay. Orphanet, which is a reference portal for information on rare diseases, defines a condition as rare when 1 in 2,000 people or fewer are affected. Parents were all over 18, were able to give informed consent and lived with their child who had a genetic condition. A member of the research team identified eligible parents from the clinical genetics records in NHS Tayside. Parents of twenty-five children were identified as potential participants. They were contacted via letter and informed of the project. One repeat letter was sent to the parents who did not reply initially. Twelve children's parents

replied agreeing to take part in the study. On receiving written confirmation that the parent had agreed to participate, they were contacted by phone to organise a suitable time and place for interview. One of the parents was subsequently unable to attend an interview.

### Interviews and Analysis

Eleven semi-structured, in depth interviews took place with parents of children suffering from ten different rare syndromes. All of the mothers and one father participated in the interviews. Seven of them took place in the Clinical Research Centre, Ninewells Hospital, and four took place in the participant's home; appropriate safety precautions were put in place. The clinical genetics team knew where and when interviews were being held and a genetic counsellor attended all of the interviews for the support and safety of the parent and the interviewer. This included the interviews that took place at home and within the Clinical Research Centre. A consent form was completed prior to the interview. Further contact with the Clinical Genetics team was offered routinely at the end of each interview. The interviews lasted approximately one hour and were digitally recorded then fully transcribed. The data source was transcripts from the interviews. A qualitative approach was employed as it allowed parent's perceptions, beliefs and emerging key issues to be explored. Analysis of the transcripts using the framework approach allowed the views of the parents' to be conceptualised in a useful way (58, 59).

The framework approach is a form of deductive analysis with 5 main stages: familiarisation with the data source to identify key themes; referencing data to the themes identified forming a comprehensive data index; adding a numerical code from the index to the text along with a description of the theme it represents; charting data according to its theme and summarising points made on each; and finally mapping which involves finding associations between different themes (58).

In this study, the children were affected by different genetic conditions. It was felt to be acceptable to compare experiences of the parents, as all of the genetic conditions were rare thus comparison between the same conditions would not be possible on a small scale. Additionally, similar members of the multidisciplinary team managed the children's cases. A

number of other research papers have also compared different genetic conditions. The themes raised in this study by these parents identified many common experiences between these families.

### Record keeping and Confidentiality

The recordings of the interviews did not contain parent or child's identification details. Full transcripts were only available to Lisa Bryson and the chief investigator. All data was anonymised and kept in password-protected computers. Quotes made are unidentifiable. Recordings of the interviews were destroyed following the completion of the study.

The questionnaires were handled in a confidential manner. No details of the consultants involved were saved.

Table 2- Details of the parents and children involved in the study

#	Stage when problems presented	Diagnosis confirmed	Age at interview	Prevalence	Type of developmental delay	Severity	*SIMD deciles
Parent 1	Few weeks old	6 years	7 years	1 in 200,000	Global	Mild to Severe	3
Parent 2	Pregnancy	8 days	11 years	1 in 10,000-25,000	Global	Mild to Moderate	6
Parent 3	Pregnancy	2.5 years	12 years	1 in 10,000-50,000	Global	Mild to Moderate	6
Parent 4	Birth	6 months	6 years	1 in 1,000	Global	Mild to Moderate	8
Parent 5	Pregnancy	2 years	8 years	1 in 1,000	Global	Mild to Moderate	8
Parent 6	15 months	6 years	6 years	1 in 10,000-20,000	Motor	Progressive	9
Parent 7	8 weeks	18 months	4 years	less than 1 in 1,000,000	Global	Severe	8
Parent 8	Birth	15 months	21 months	1 in 1,000-5,000	Global	Mild	8
Parent 9	Birth	4 months	3 years	1 in 10,000-25,000	Global	Mild to Moderate	7
Parent 10	Birth	16 months	8 years	1 in 2,000-20,000	Global	Severe	7
Parent 11	Pregnancy (12 weeks)	Pregnancy (20 weeks)	7 months	1 in 3,300-56,000	Motor	Mild	7
Parent 12	Birth	10 months	22 months	1 in 7,500-20,000	Global	Moderate	1

#

\*SIMD- Scottish Index of Multiple Deprivation

## Results

Four main themes were identified from the transcripts: the reaction to having a genetic diagnosis; how they felt the healthcare team managed them; the support that was available and how a genetic diagnosis affects the family. These themes were further divided into subthemes as shown below. The themes and subthemes can overlap and link to others.

The reaction to having a genetic diagnosis included:

- Parents' experiences of not knowing the diagnosis
- Receiving the diagnosis
- The benefits and drawbacks of having a diagnosis
- The emotional reaction to the diagnosis
- Living in a "bubble"
- Coping mechanisms

How they felt the healthcare team managed the family included:

- Hospital appointments and healthcare professionals
- Meeting the parents' emotional needs
- The impact of the rarity of the condition on hospital management

The support available for families included:

- Introduction into a "new world" and support required
- Financial support and housing
- Information
- Using the Internet
- Where the parents found support
- Education and Schooling
- Respite care
- Family and friends as support

How a genetic diagnosis affects the family included:

- The impact of conditions being genetic
- Family relationships
- Friendships
- Family planning



## The reaction to having a genetic diagnosis

### Parents' experiences of not knowing the diagnosis

During the interviews the parents were asked about how they felt gaining a diagnosis for their children suffering from a variety of different genetic conditions causing developmental delay. It was unanimous that having a genetic diagnosis was better than not knowing what their child was suffering from. A great deal of worry appeared to be associated with the unknown. Mother 9 compared her situation to her friends' who did not know the diagnosis for their child with a learning disability. She described her friends' family as being in "limbo" because they did not know what was to come or how to plan for the future. She appeared glad to not be in their position. Mother 3 described automatically thinking the worst before her child's diagnosis had been found. She believed that the condition which her child had may be life limiting and this belief affected the development of their relationship.

"I was a bit standoffish, because I just didn't know, that sounds awful as a mother, we did bond, but I kind of felt I don't know how long I'm going to have her. I don't, you just didn't want to." - Mother 3

Some parents gave an explanation as to why they felt having a diagnosis was beneficial. Feelings of guilt were mentioned in relation to not knowing the origin of the condition that their child suffered from. Mother 5 had concerns about problems in pregnancy and during delivery. She felt something must have gone wrong at the time that she had not noticed. Mother 7 had suffered from epilepsy and was concerned that the problems, which her child had, were related to her condition. When finding out the true diagnosis, this feeling of guilt was relieved, as the mutation was *de novo*.

Mother 11 provided a different explanation to the benefits of having a diagnosis. She was concerned about her child, as she was a twin, which made it "obvious" that there was something wrong. The comparison between the twins highlighted the developmental delay. Without having a diagnosis, she would have no explanation as to why there was a difference.

Some experienced mothers explained knowing immediately that something “was not right” with their child, whereas newer mothers described not recognising any signs of a medical condition. For mother 4 not knowing the diagnosis was particularly upsetting as she felt it lead to her parenting skills being questioned. She was sent on a parenting course as her two youngest children were developmentally delayed. She felt she knew what she was doing as her eldest was developing well. She described her lack of skills to provide for her child being blamed for her children’s problems before the real diagnosis was known.

While waiting for a diagnosis, some doctors provided information on possible diagnoses. Parents discussed their experiences of this. Mother 10 was told that her child could have Edward’s syndrome whilst she was pregnant. She found this upsetting and unhelpful, especially as it was not the correct diagnosis. The true diagnosis was not life limiting and was less severe.

At the time of interview, mother 10 did not have genetic confirmation of the condition, as there was not a genetic test available for it. She felt having genetic confirmation would only be beneficial if it would alter the management of her daughter’s condition. Mother 6 also did not yet have genetic confirmation of the diagnosis as the gene mutation causing the condition had recently been identified. Her child was undergoing further genetic testing during the interview. She discussed how it would feel to know the diagnosis was molecular rather than clinical.

“It will be good to know for definite. I never thought they’d ever identify the gene that caused it. I just thought that it would be, “oh well he’s probably got that” and that is fine. But for them to actually have identified the gene is quite good and to have a definite, 100% diagnosis.” - Mother 6

### Receiving the Diagnosis

Recalling parent's memories of diagnosis caused some emotional responses. 5 of the mothers became tearful while remembering this time.

All of the parents described reacting slightly differently to the diagnoses. Mother and father 9 explained having very different reactions to the same condition. The father explained feeling he had to be a strong male, remain positive and get on with it, whereas the mother had a pessimistic view on their situation and became emotional.

Some of the parents had been waiting years for a diagnosis. Mother 6 said she knew there would be a diagnosis. She felt this made the event of receiving a diagnosis inevitable and perhaps unsurprising. The management was already in place so the diagnosis did not alter the care of her child.

"There wasn't really any change. When we got the diagnosis, it wasn't anything that exciting really. It was alright to have a name." - Mother 6

This is the opposite of how both mother and father 9 felt. They thought they had waited such a long time to find out whether the result was positive that they began to assume that everything was fine. They assumed the results were negative, as they had not heard otherwise. This made receiving a diagnosis surprising and upsetting.

"I was really upset to begin with because obviously I'd gone into that appointment thinking that everything was fine, it had been ages, if there had been anything wrong we would have heard back now type of thing." - Mother 9

Mother 11 was given the idea that they were checking for genetic mutations but that it was extremely unlikely that anything would be found. Thus she was also surprised when she found out the results had returned as positive. She felt happy with how she was managed at this stage.

Some parents discussed breaking bad news and what they found to be helpful and unhelpful. Mother 5 had been involved in training NHS staff in breaking bad news due to her bad and good experiences. She had been given two diagnoses for her child, the first was

Cerebral Palsy and the second was her child's true diagnosis of a degenerative genetic condition.

"It was absolutely dreadful because this guy came in and said "oh, of course you know he's got Cerebral Palsy don't you" and then I was left alone. Everyone left. I was given no information, I was given no follow up, I was given nobody to talk to or anything. It was just so lonely, so overwhelmed and I had no idea."- Mother 5

She compared this example of breaking bad news to her other, more positive experience. She was told her child had a genetic condition by a paediatrician that she had a good rapport with. The paediatrician knew her family background and could empathise with her concerns for her other child. She was told face to face and given time to voice her emotions. She had positive memories of this experience.

The type of diagnosis that was being given appeared to have an effect on some of the parents' reactions. For example, mother 8 had a daughter who had been diagnosed with autism and a son who had been diagnosed with a genetic condition. She compared getting each of these diagnoses. She felt autism was more difficult to come to terms with as she thought it was a diagnosis based on a doctor's opinion, whereas having a genetic condition could not be disputed.

A few of the parents mentioned the earlier the diagnosis is received the better. Mother 10 appreciated finding out the diagnosis of her unborn child in pregnancy, as it meant she could speak to families affected by the condition and watch a TV programme on it before her child was even born. She found this reassuring. Mother 2 was given the diagnosis within a few days of birth. She found this helpful, as she found the period of uncertainty unpleasant.

"If I was just told there was a problem then you're going to end up worrying even more ... to be given a diagnosis so quickly was a huge relief."- Mother 2

### Perceived benefits and drawbacks of having a diagnosis

Receiving a diagnosis was seen as beneficial by all of the parents. There were many different reasons given to explain why this was the case. Mother 3 described being reassured by the diagnosis, gaining access to further information and support and being able to plan their family's future more easily.

"Right away, we went home that day having spoken to the geneticist and we looked it up online and there was a picture of a child and that was her. She had the facial features everything. And we went from there, joined the support groups, got a book on it, looking at research papers...we had a rough idea as to what to expect and it was better than we thought she might have had." - Mother 3

Mother 4 discussed the importance of gaining confirmation that the concerns she had about her child were justified. She felt she was not being taken seriously and was being treated as an over protective mother until they gained a diagnosis. She wanted to know that the delay in her children's development was not due to her parenting.

Mothers 3 and 10 worried about more serious conditions and therefore receiving a diagnosis was beneficial as it provided the family with hope for the future. Mother 3, in particular, noted her concerns that her child's condition was "life limiting". She felt this affected the development of their relationship. In contrast to this, the diagnosis may be negative if the prognosis for their child was worse than expected. Mother 8 described gaining a definite diagnosis resulting in the realisation that their child would not grow out of the problems they were having. This caused her to lose hope.

"It is a rubber stamp that your child is always going to have special needs. There is that kind of definite root. Before you always kind of had that hope that it might just be a temporary blip and when he's 1 he'll catch up." - Mother 8

Some parents found having a label for the condition their child was suffering from helpful and some did not. Mother 9 felt the diagnosis was confusing and she was not provided enough information to understand what it meant. She described the diagnosis as being "a name floating". Mother 11 liked having a label to explain to her friends and family what the

condition is. She describes her child's syndrome to friends by explaining "it's kind of like Down's syndrome".

Having the label also led to more questions from friends, families and others as the name meant very little to people. Mother 9 explained that telling someone that a child had a rare diagnosis is not like saying Down's syndrome, which is a condition that most people can understand in her opinion. Mother 6 felt having the diagnosis did not provide a useful label as she also felt people did not understand it. Additionally, she felt having the name did not benefit healthcare professionals, as they also knew nothing about the condition.

"Nobody knows anything about the syndrome so it doesn't make any difference really."- Mother 6

Many of the parents also voiced concerns that the label would define their child. Mother 3 talked about her siblings being very different. One of them does not have a genetic condition but was still very different in many ways as she described everyone having their own differences. Mother 10 also voiced concerns about her daughter becoming defined by her condition.

"I'm more concerned for her, I don't want her to grow up thinking that's all there is to her"- Mother 10

The genetic aspect of the diagnosis caused many different emotions in the parents. Guilt was mentioned in relation to the origin of the gene. Some parents mentioned blame being placed on certain family members. Mother 5 mentioned the fear that she was suffering from a late onset form of the same condition as her son. She was concerned that she could end up in a wheel chair or could have passed the faulty gene onto her other child. This caused her to feel mixed emotions, which she seemed to find confusing.

"It's quite funny you feel really guilty that I really don't want my daughter to have it but my son has it and it's really, you just feel that you're not wanting to take or detract from the child that has the disability, you know have the disorder but I really didn't want my daughter to have it."- Mother 5

### Emotional Reaction to the Diagnosis

Many parents discussed the grief reaction related to the diagnosis. Some parents gave their reasoning behind why they felt they were grieving. Mother 5 discussed grieving as their child was going to have to suffer more than they deserved because of the diagnosis. Mother 10 grieved because her child was not going to be able to join in normal activities, which other children enjoy. Mother 4 grieved for the child she thought she was having and had lost.

Some parents received 2 diagnoses, the first of these being incorrect, which lead to a second grief reaction. Mother 10 was told while she was pregnant that her child had Edward's syndrome. She found this extremely difficult, so when she received the true diagnosis with a much better outlook and prognosis, she felt relieved. While mother 6 was given a worse prognosis which she felt was frightening, although she had already come to terms with the first diagnosis so it was easier to deal with the second one. Mother 5, who had her child diagnosed with Cerebral Palsy prior to getting the true diagnosis, mentioned that she may have found it easier to deal with the grief second time around as she know what was coming.

Postnatal depression was discussed by 3 of the mothers. Mother 3, who discussed being surrounded by professionals at different clinics and appointments, mentioned that it was not until she was back at work that her workmates realised she had postnatal depression. Mother 2 related her postnatal depression directly to her worries about the early diagnosis of a genetic condition in her child.

"Thinking back now I really genuinely think that had child 2 had no problems and been a normal baby then I don't think I would have, I can't see why I would have been. I think it was because of all the worry and concern about the condition and that's what did bring me down." - Mother 2

Mother 3 discussed her ex-husband and his difficulty in dealing with their daughter's condition and the amount of time they had to spend in hospital "he got quite depressed and ended up he was suicidal". She described him then requiring psychiatric inpatient care. This added to her stress levels and as a result she found it difficult to cope.

In contrast to this, mother 7 did not feel she had a grief reaction to the diagnosis. She discussed being positive as his disabilities were motor and not global. She felt this was easier to deal with, as he did not have an obvious learning disability.

“One of the main reasons why I would say it doesn’t worry me too much is because he is incredibly bright. His speech is, he keeps up amazingly with his speech” - Mother 7

Additionally, mother 7 described herself as feeling lucky to have the opportunity to simply have a child. She did not describe feeling grief towards the diagnosis. Mother 6 also felt lucky to have a child with such a rare condition. It made her feel special. Mother 4 described her grief for the child she thought she was going to have and how she came to terms with it by reassessing her situation and realising how lucky she was to have such special children and watch them overcome their obstacles.

“Then you go through a process of like grief really.... you almost grieve for the child that you thought that you gave birth to but then you kind of regroup and reassess and realise they are still you’re children, it’s nothing they’ve done. They’re still wonderful gorgeous people” - Mother 4

### Living in a “bubble”

The word “bubble” was mentioned by many of the parents. The ways in which the parents used it appeared to differ slightly.

Mother 8 talked about being in a bubble that prevented her from living a normal life, as she only had time to focus on her child. Mothers 1 and 11 felt they were in a bubble as they were only able to think about what was happening presently and were not able to plan for the future. Mother 11 felt too much had happened in the past and felt it was a waste of time trying to plan for the future.

“Try not to discuss what’s going on unless it’s to the forefront. And it’s on the calendar. It’s just our way of dealing with it.” - Mother 1



Mother 8 described feeling similarly to mothers 1 and 11. She feared that suddenly things were going to come crashing down on her, as she would keep going and only think about necessities. She felt that she put up a front and acted like she was doing well to the health professionals as a way to protect herself. She speculated whether this might have caused her to have less access to support.

Mother 5, who also described herself as living in a bubble, explained feeling happy to discuss the condition, which her son suffered from, and understand what it meant for the future. Thus, she explained how surprised she was at her own reaction to reading a letter from her doctor on what they had previously discussed.

"We got a letter from child 5's orthopaedic consultant that he saw in clinic a couple of weeks ago. And it said child 5 is deteriorating full stop. And you read it and we know we had that discussion and it was absolutely spot on ... but actually when read it I just went oh no I don't want to see that. ...Yeah you get on with it and then you see it and go AH!" - Mother 5

Some parents talked about the journey that they were on in order to come to terms with the diagnosis. Mother and father 9 and mother 10 felt it was attending conferences and being in contact with others in same position as themselves that helped them out of the "bubble". Seeing others at different stages in life allowed them to think about the future and realise that other families were managing. The mother and father 9 felt they were no longer denying their problems but realised that they had been prior to attending their first conference. They described living in a bubble and thinking about life on a day-to-day basis. They similarly to above did not think about the future. Mother 10 had gone to a conference expecting to be in a bubble. She thought at the conference she would realise how bad things would be. However, it had the opposite effect.

"I think I was worried that I was just in a bubble and then it would hit me that I would see all these poor children and think oh my god, the future is bleak...but I don't know why, I mean it was great going there, it was good, it was really reassuring." - Mother 10

### Coping Mechanisms

Different ways of coping were described and pulled out from the interviews. Mother 6 explained the diagnosis was “one of those things that’s happened... You’ve just got to get on with it”.

One coping mechanism, which was used by mothers 5 and 8, was focusing all of their energy on the child to ensure their full potential could be achieved with the support available. These parents discussed how organised and professional they had to become due to the number of appointments and the amount of care their child required. Mother 5 described herself as acting as the “project manager” of her child’s care. She was pleased when her child told her he was happy with who he was. Mother 8 discussed her child also having normal needs, amongst the professionals and appointments, which had to be remembered.

All of these parents had opted to take part in this research study. Being involved in research was a coping mechanism, which some parents identified. Mothers 3, 5 and 10 in particular discussed taking part in a large number of different studies. This gave them something to keep their minds occupied.

“There’s always research on going into this. And we’ve just participated in another one, but that was all about psychological problems that they have.” - Mother 3

A number of the mothers described the positive experiences that they had raising children with additional needs. This outlook appeared to help them cope. Mother 2 had imagined that her child’s delay would be more significant than it was, so when her daughter was able to go to mainstream school there was a real sense of achievement. Mother 4 described how fulfilling it was to have a child with a disability who has to overcome obstacles which other children do not have to overcome and take for granted. This made her feel proud. Additionally, mother 4 described the condition as just being a part of who her children were and therefore she did not think about the diagnosis with any negativity.

“They are all the more special for their difficulties...Just seeing how they overcome everyday challenges that most children just take for granted, you know being able to talk or run

around with their friends and everything else. It's a humbling experience; it teaches you a lot about yourself." - Mother 4

Mothers 5 and 9 discussed comparing themselves and their own situation to other families whom they felt were in a worse position. Comparing their child's disability to other children's disabilities meant that they felt thankful for what their child was able to do. They felt seeing other families managing helped to put their own problems in perspective.

Parent's general outlook on life appeared to affect how well parents coped. Mother 7 described herself as being laid back and positive. She did not feel stressed by the diagnosis and felt she was coping well at the time of the interview. Mother 6 compared her outlook on life to a friend's. She described herself as being quite scientific and positively minded. She found it easy not to get stressed, whereas her friend who had a more negative outlook on life had problems with depression. Mother 6 additionally hoped that as the condition varied between children, her child might not be affected as severely as all the others have been.

"Of the 13 children that they did the study on, 10 of them had died before they got to the age of 5. So I don't know what that means if it's classed as life limiting or not" - Mother 6

In relation to coping with the diagnosis, parents' personal health, family and educational backgrounds were thought to be a relevant factor. Mother 6's sister was a doctor, who was able to help her understand information and research the condition. Mothers 4 and 5 both had degrees in biology and felt they found it easier to understand the diagnoses and the terminology used by doctors. They both felt they benefited from this.

Similarly, parents' health affected how they coped with the diagnosis. Mother 11 felt they were laid back about the diagnosis as her partner had Crohns, which he was in and out of hospital with already. Mother 7 was also not concerned by her son's condition and related her laid back approach to having epilepsy. She felt as long as her child was happy she did not have concerns.

"I'm fairly laid back about it all. I've always worked on the fact that he is very happy and so I just think as long as I've got a happy child then I'm not too concerned...It's probably because of the way my life has been that I'm so laid back about this, because of my epilepsy and

because I was lucky enough to have an operation 3 years ago that has made me as well as I am now." - Mother 7

Not thinking about the future helped some families cope. Mother 3 felt that due to the amount of variety within the particular conditions, planning for the future would not be feasible and so they should make life as normal as possible. Mother 7 felt you have to get on with it and not think about the future because it would be a waste of time to worry when your child is young. In contrast to this, mothers 2 and 4 put a great deal of thought into the condition and how to cope in the future. Mother 2 had started saving money for her child's future and mother 4 was thinking about who would be a carer for her children in the future.

"We'll be getting older and who'll be able to help her when we're not basically you know further down the line... we have started saving money away every month for the future, for whatever the reason we might need it for, we had money going away every month." - Mother 2

## How the healthcare team managed the family

### Hospital appointments and healthcare professionals

There were many complimentary statements made towards the healthcare team in general. One parent in particular was very appreciative of the management they had received and could not think of anything she would have changed.

"I really couldn't complain about anything at all just now, I'm very happy with the way it has all been dealt with. I'm a big supporter of the NHS. Being as lucky as I am, I am a huge supporter of it." - Mother 7

Father 9 talked about "The Team Around the Child meetings", which involved the multidisciplinary team gathering to discuss the child's needs and a management plan for each of these. He described this as "helpful".

However, there were a number of other comments, which suggested that the management of these families could be improved.

"You've got all these people to see, you've got all these appointments" - Mother 9

"I'm not going to sit and explain child 1's whole life all over again" - Mother 1

Many of the parents discussed the number of hospital appointments and healthcare professionals involved in their child's management. Mother 9 explained that out of 5 working days, she could remember having 4 days taken up by appointments. They lived rurally making travelling to hospital appointments very difficult. Mother 1 discussed her other child feeling left out due to the amount of time and attention that was required in attending hospital. The number of clinics deterred mother 1 from seeing healthcare professionals altogether. She decided to stop going to clinic appointments as she felt they were not of value for her or her daughter. She felt her daughter was spending too much time in hospital and not enough time living life.

"I made the decision that we need to get off the wheel. And she needs to live a wee bit of life...Your whole life is run by people saying come to this clinic and come to that clinic, I just took control and said you're not doing anything for us and we need to get off" - Mother 1

Additionally, due to the large number of people involved, it was mentioned that the child's story needed to be repeated numerous times, which could be upsetting. Mother and father 9 discussed doctors looking up the condition online during clinic appointments or asking them again to explain what the condition meant. Mother 1 described how she felt about seeing different consultants who did not understand her child's history.

"I was very much getting angry at every clinic that we went to. And we were seeing a different consultant. I was like I don't want to see you. I'm really sorry but I see the consultant that I was already seeing because I'm not going to sit and explain child 1's whole life all over again." - Mother 1

The amount of people that parents saw caused confusion about who they were and what their roles were. Mother 5 explained getting mixed up between "orthopaedic consultant and an orthotist and an occupational therapist". She did not know who was who and who could provide her with the help that was required.

Some parents stated that having one constant supportive team member, who could help them to manage their appointments in a more time productive way, would be beneficial. Mother 9 wanted someone to organise the appointments so they could all be within one day. Then the rest of the week she could enjoy time with her child. She felt nobody was taking on board how difficult it was with a small child to travel for appointments numerous times every week. Mother 11 had a Parent-to-Parent support worker who she was close to and felt supported by. She felt she could talk to her and ask her about anything and she would receive advice, which was valuable and specific to her and her situation.

"She is just my key worker now and she does everything, any queries I have I go through her" - Mother 11

### Meeting the parents' emotional needs

Certain parents discussed feeling supported in a clinical sense but emotionally their needs were not always taken into account. Mother 8 felt she would have liked more support from the healthcare team in particular.

"The only thing for me that could have just made the experience a little bit more sort of bearable is if there had sort of been somebody who you could actually have a proper conversation with and not feel as though their time was. And have that somebody there on a regular basis"- Mother 8

Communication and breaking bad news were areas of hospital management, which were highlighted by some parents, as requiring improvement. Mother 2 recalled healthcare team members not treating them with as much empathy as they would have liked. They felt this was due to staff being too rushed to provide the kind of support that they needed.

"I don't think she had a lot of bed side manner skills to put it kind of as nicely as possible. The way she did it, it was all done in a rush, and she was kind of hassled and it just wasn't the best situation"- Mother 2

When being given a diagnosis, some parents discussed the need for further follow up, for example a phone call a few weeks later to make sure things had sunk in, an appointment to discuss what they had read and what they perhaps did not understand. Mother 3 described being told something was wrong in pregnancy. She was given the information and asked if she wanted to terminate. Then when she decided to continue with the pregnancy, she was given no further support or follow up. She felt she needed someone to speak to who would listen to what she was going through. She said this was not available to her. Mother 4 also requested a need for counselling after diagnosis to talk about how she felt about her situation.

In relation to this, at the time of diagnosis, mother 11 was advised to go home and let the information sink in, then return for an appointment to discuss any questions that they have. She thought this was good, as she did not know what to ask at the time of diagnosis. She made a list of questions, which she would have liked to ask. However, the appointment was

lost and there was a year between diagnosis and follow-up appointment. Mother 11 felt this was far too long as by that time she had an understanding of the condition from her own research and living with a child with the condition. She felt she needed follow up 2 weeks later. She felt if the doctor could not see her, then an appointment with a "liaison officer", who was able to explain the diagnosis and answer questions, would have been helpful.

Mother 8 wanted someone to come to appointments with her which she could "bounce off of and kind of say 'that bit there, did I hear that bit right' other than my husband". Mother 6 also wanted follow up after receiving the diagnosis. She did not receive a further doctor's appointment either.

"It would have been good to be able to talk to somebody else about it after I had then gone and done a bit of research... I suppose it might have been good after I had got the diagnosis, to get a phone call from somebody a month later to say how are you coping with it? How are you finding everything now?" – Mother 6

Waiting for test results was reported as being a very stressful period for families. Mother 5 would have appreciated an appointment to receive her own test results. She was told instead to wait for a phone call in which they would inform her whether she was a carrier or not. The stress of waiting for test results was exacerbated by not having a time to receive them. Then when the results were back and no one contacted her, she tried for days before managing to get through to anybody. Additionally, she pointed out that had her results been positive she would have wanted to discuss this with someone face to face and have her husband by her side for support.

Emotional problems caused by waiting for test results appeared to be a common theme. Mother and father 9 stated that they had waited so long for test results that they started to assume all the results were negative and felt relief. Then receiving the diagnosis after a year was even more of a shock. They felt they should have been kept informed with updates. Another cause of emotional distress was doctors providing incorrect diagnoses before the test results had returned. Mother 10 in particular was told her child had Edwards syndrome incorrectly when the true diagnosis was much more benign.



A fall down in management, which was highlighted, was a lack of good signposting to support, information and details on what to do next. Parents reported having to find information on their own and support was not provided or was provided in a staggered disorganised approach. Mother 4 suggested that providing a checklist for parents could be helpful as a more “consolidated approach” to support was necessary. She felt being offered and finding support was down to luck.

“Maybe there needs to be a sort of, almost a sort of checklist to go through. Have you been in touch with the relevant support organisation and have you been offered counselling.”- Mother 4

### The impact of the rarity of the condition on hospital management

The rarity of the conditions that were included in the study had an impact on the hospital management that the parents received.

Many parents discussed feeling like the healthcare professionals managing them did not understand the condition due to the rarity. It may be the only case the healthcare professional has seen. Mother 11 felt “My doctors don’t have the slightest Scooby”. Mother 8 discussed having doubts about what she was being told at clinic appointments because she felt she understood the condition better than most of the healthcare team. Mother 6 who had a child with an extremely rare condition understood the lack of knowledge relating to her child’s condition.

“Because it’s such a rare syndrome, nobody’s ever met another child with the same thing. So it’s not like they can say, “well in my experience of children with this syndrome we do this”- Mother 6

On the other hand, some healthcare professionals did read about the conditions and the parents were grateful to speak to someone who understood more about their child’s condition than they do. Mother 3 discussed understanding her child’s condition was rare so all the research that she reads cannot be read by the healthcare team. However, when one

of the registrars took the time to look up her child's condition and ensure management was optimal, she felt reassured that there was someone in Tayside who was "on the ball".

Parents reported gaining a better understanding of the condition that their child suffered from than the healthcare team. This was seen as positive for some parents as they were able to take control of their management. However, this also led to disagreements between the healthcare team and the parent on how the child should be managed. Mother 8 expressed a need to have a conversation with a doctor who had studied and read the research. She felt different therapies and management were available but doctors were not explaining to her why her child was not being given them when she asked.

"There is no explanation as to why your child can't go on that hormone or why that is not. There is no real dialogue or conversation and I think I know that a couple of doctors are slightly frustrated with me because of the amount of reading that I do about the syndrome"  
– Mother 8

Some parents attended clinics that were specialised for the condition that their child was suffering from. These clinics were not in the Tayside area. Parents felt they were helpful as the doctor knew and understood the condition and everything could be checked at once. All areas important for the certain syndrome could be covered within a day. Additionally, mother 8, who attended a specific clinic in Glasgow, stated that they would be willing to travel to London for the clinic, as it was so beneficial to talk to specialists who understood the condition. The parents who did attend these clinics tended to discover their existence through parents they met from the organisations and then they would request a referral. Mother 4 heard about a clinic in England and discussed how beneficial it would be if she were to have access to one similar. She felt she wanted to send a letter to try and rally up support for getting a 1-stop clinic in Scotland for the condition her children suffer from. Mother 11, on the other hand, discussed attempts made by different team members to attend clinics together. She felt this did not work, as she needed time to think about her child's problems separately.

Due to the rarity of some of these conditions, some parents felt that their child was of interest to medical professionals. Mother 1 felt that their child was treated like a "freak" as

people wanted to see her. Mother 10 talked about having students in the room. She did not feel she was given the opportunity to provide consent.

"There were a couple of times that I was asked when (the students) were in the room and I just think that's not consent. I am coerced into saying yes. I'm emotional, I'm fragile, don't ask me when they're in the room"- Mother 10

All of these areas highlight a need for the management of these families to be considered further. None of the parents reported a huge change in management with the diagnosis. This suggests that it is possible to ensure children are managed fully without having a diagnosis.

## The support available for families

### Introduction to a “new world” and support required

“Very quickly you’re kind of, you’re just completely thrust into this world”- Mother 8

Parents reported that in the early days, with or without a diagnosis, they can feel lost in the management and not know what to do or where to go. Many of the parents compared this transition to having a child with additional needs as like being introduced into a different world. It was found that parents gained an understanding over time and often felt able to manage their child’s case as they became more knowledgeable about the condition, gained an understanding of the child’s needs and what management and support was available to them. Mother 5 described how she felt when her child was given the initial diagnosis of cerebral palsy.

“You are just thrown into this world, it’s like you go through a time warp and go into hospitals and you see all these people ... years go by, you get to know and you get to know the people and you get to build relationships up with people and that would really work.”- Mother 5

Parents described being “thrown into this world” at different stages of their child’s life, with or without the true diagnosis. In some cases, parents had been in the disability world for years before they got a diagnosis and this relates to parents feelings that the diagnosis had no effect on the child’s management and support. Mother 5 felt that once she had an understanding of how to manage her child’s needs and felt less lost, she realised “it’s a world you go into and you embrace and actually even get quite a lot of fulfilment out of it.”

As well as parents feeling lost, some also described having to fight for the resources that are available to them. Parents discussed feeling that they need to continually prove that their child has a disability. For example, mother and father 9 described fighting for the level of disability allowance they deserved. Furthermore, mother 8 described being watched when parking in the disabled bay at the supermarket.

“He’s got a blue badge and then you go in a parking space and you get people looking at you like you’re not disabled, and you’re kind of like actually my child is. But he is in a buggy.

Yeah, but that's because he fits into 12-18 month clothes and I want him in a buggy, maybe in the next year you'll see him in a wheel chair. But why should I have to justify why my child is in a buggy." - Mother 8

The parents who were interviewed described concerns that they had which most other parents would not have to worry about. Mother 5 talked about having to come to terms with wheelchairs that no longer are an issue for her. It took time to accept that her child would require one. Mother 10 talked about having to consider what dinner forks were suitable and making light switches more accessible for her child's disability. There were "home-made gadgets and contraptions" which she had been told about by other parents to make her child's life easier. Mother 4 described having to take sign language courses to enable her to communicate with her children.

Parents felt that their lives changed significantly with a child with a disability. Mother 6 said her "social life it is practically non-existent". She said that this did not bother her as she enjoyed spending time at home with her child. Other parents did not view this problem as positively. Mother 8 felt all of her old friends were living different lives that she was unable to be a part of.

"What they are getting up to and all that and to a certain degree probably life, has bypassed because we are in this bubble. So yeah it is quite difficult. But like I say life goes on and you've got to kind of just get on with it I suppose. ... I'm sort of fine with it almost because there is no sort of alternative" - Mother 8

Holidays were also affected as a result of the child's needs. Mother 4 described their most recent family holiday. She organised the holiday "with a certain holiday company who pride themselves on having a very good child care facilities". Yet they would not listen to what she said when she was describing her child's needs. They refused to allow her youngest son to attend the children's clubs due to his medical problems. Mother 4 was upset about this as he usually attended mainstream school. She felt her son was being discriminated against.

"They are a British company and they are employing British nannies and they should come under or comply with... the equality act, however because the service is being delivered in Portugal they didn't have to comply. So, that was an interesting one" - Mother 4

Having all of these things to think about can have an impact on siblings who may miss out on holidays and activities that other children take for granted. Mother 3 discussed her other child being bullied as a result of having a sibling with additional needs. Also, many parents talked about how they hoped the sibling would take on the care of their sibling with additional needs when they get older.

Many of the parents discussed their concerns about the future. Mother 7 discussed being concerned that her son may require a wheelchair one day but she did not want to think about it too much until they knew what was going to happen. Mother 10 discussed being at a gala day and seeing “little girls dressed up and highland dancing” and she realised her daughter would never be able to join in and felt quite upset at that thought. Mother 2 felt it was important for her daughter to find a role in life but was concerned that she will “struggle to live an independent life” and as they, the parents, get older, they will be less able to support her. Mother 4 did not want to think about the condition her sons suffered from with any negativity. As the sons got older she watched them “overcome everyday challenges that most children just take for granted”. She described this as “humbling”.

Parents described the many added difficulties that their families face and may not be considered by the team supporting them.

### Financial support and Housing

Parents’ lives were affected in many ways by having a child with additional needs. Parents reported changes being made to their working life. For example, mother 9 stopped working altogether. She related this to the number of hospital appointments and the amount of care necessary to look after a child with additional needs. Mother 8 also described herself as being “unemployable”.

“There was nobody that was going to employ me and I’ve never worked since, which has been a bit crap”- Mother 9.

Some parents had more understanding employers and were allowed more flexible hours. Mother 3 worked part time and felt this did not affect her much as benefits could make up

the difference in her income. However, as a result of job changes, many of the parents described the income of their families decreasing.

All of the families involved in the study were eligible to claim benefits. A number of the parents reported not realising they were initially. Mothers 1 and 2 did not realise for years. Mother 1 felt the healthcare team should have told her about this and not her work colleague. A health visitor asked mother 7 if she was accessing benefits prior to the interview. She was also unaware that she was entitled to financial support.

“Are you not getting disability living allowance?” I thought I won’t get that, she said don’t be daft you’ve got a named disorder, she will get. I didn’t know anything about that... I remember thinking at no point had anybody in the hospital” - Mother 1

Mother and father 9 described being turned down for benefits before having a diagnosis and once they had a diagnosis they had to “fight” for the level of benefits that they were eligible for. Mother 11 described the help that she was given by her Parent to Parent support worker. She helped her to complete the forms correctly and organise the correct financial support for which she was entitled.

“Parent to parent support worker so she comes in quite often and when she first came in she made sure that child 11 was getting all the benefits she was due. This year we’ve booked a holiday and she’s actually asked for funding for us for the holiday because child 11 is entitled to it.” - Mother 11

Parents may opt to use their benefits in different ways depending on their financial situation. For example, mother 2 started saving it for her child’s future, as they did not know if she would be able to live an independent life. Mother 4 used the benefits for help in the house, caring for her two children who were not affected by the condition, whilst she took the two that were affected to their hospital appointments.

In relation to above, parents explained how expensive it could be to have a child with additional needs. Mother 5 described the amount of petrol required for the number of hospital trips and the specialist equipment that was needed. All of these extra expenditures

may be difficult to cover and fundraising may be required. Father 9 joked “A winning lottery ticket that would do”.

Housing in which the family live may not be appropriate as a result of the disabilities from which their child suffers. Due to money problems there may be limited options available to them. Mothers 8 and 9 described not being able to make the adaptations that were necessary for their children due to a lack of funding. Both parents discussed the likelihood of having to sell their properties as a result, which they both felt would be frustrating.

“I’m quite proud of the fact that I’ve had a mortgage since I was 21 years old and I’ve worked really hard for it and it is quite frustrating and upsetting to think that I could lose all that.” - Mother 8

### Information

The conditions from which the children suffered were rare and therefore the parents did not automatically understand the meaning behind the name. When given the diagnosis, information was required to explain what the condition was and what it meant for them. However, many parents had complaints about the information that was provided. Many of the parents understood information on the condition might be limited due to its rarity and it might be difficult for the doctor or healthcare professional to provide information as they might not know a lot about the condition or have ever had to provide information to a family that is affected. In spite of this, many parents want doctors to know more so that they can explain the conditions properly and provide reliable information and answers to the questions they have.

Some parents reported being given very little information with limited direction to reliable resources. Mother and father 9 said they were given one sheet of A4 printed from the Internet. Mothers 6 and 11 said they were given nothing and had to look it up themselves. Mother 11 stated that because she was not given any information home with her or advice on where to look she “automatically look(ed) on the Internet and on the Internet you automatically find the worst case scenario every time”. Many others reported the opposite.



They were given a hand out of information that gave too much detail and often worse case scenarios. Some felt that the doctor had not read or considered what they were handing over. Mother 2 felt the pile of information she was provided at diagnosis was inappropriate at that time. She felt in "shock" at what she had been given. Mother 3 was similarly provided with too much information that she felt made her "demented". She thought the doctor might have given her more information than usual as she was a trained nurse.

Many parents noted that they found the genetic information difficult. They could not remember what they had been told and would become confused. It was mentioned a number of times that the information doctors gave may be overly clinical and may not relate to the specific family and what they want to know. Mother 5 remembered being told about "the locus of blah blah blah" when she really wanted to know how it was going to affect her family. Mother 10 remembered hearing about "micro RA something, some complicated thing with all the genes". Mother 1 felt similarly about the genetic information she was provided with.

"It kind of all went around in a bit of a mush of yes she has this gene but what did it actually mean?" - Mother 1

A few felt the information had been oversimplified and this might fail to portray the true meaning of the child's condition. It was suggested that doctors might not provide accurate information and suggest that everything is fine when it is not. They may be rushed and therefore not give parents the time required to understand what the parents need and want to know. Mother 2 explained her experience with a doctor whom she felt did not give her accurate information.

"She was going to struggle to thrive for the first sort of 6 months to a year. And then things should get a lot easier. Now that's maybe part of the story but that's the wrong thing to say. It made me think you know after 6 months everything will be fine." - Mother 2

### Using the Internet

"The Internet is wonderful and not so wonderful"- Mother 10

The Internet was noted as a resource of information that could be very negative and unhelpful. Some parents were advised to do research and some were advised not to. Mother 7 was advised not to, which she felt was good advice. Many parents have negative experiences of searching blindly online without direction. Additionally, some parents found it difficult to know whether information was relevant to them. Many of the syndromes can vary in the way they present and how the children are affected. This made mother 1 confused about the information that she found on the Internet.

"It's got the same name but does it mean the same thing"- Mother 1

Many parents highlighted how unhelpful some American websites could be, as they seemed to focus more on the extreme cases, whereas UK sites focused on general problems. Websites from other countries may also discuss management and resources that are not available within the UK. Mother 2 had experience of reading the "worst case scenarios" on American websites; she described it as being "horrific". She felt the British site was more relevant to her.

The Internet can also provide areas of support and certain websites were very helpful. In particular, Facebook has been noted as being a very valuable resource. It allowed parents to speak on line to others affected by the condition. They could discuss problems they were having and be given advice quickly from people who have gone through the same thing. They can do this from the comfort of their own home without having to meet people face to face. This allowed for a safe place to get information without seeming vulnerable in person. Mother 6 in particular discovered that the gene test had become available for her child through a friend on Facebook from Holland. This information was available to her before her doctors had heard about it. Mother 11 felt that Facebook provided her with the most relevant personal information. Mother 9 also had a lot of positive comments to make about Facebook.

"Facebook is amazing. Just it's like this wee family"-Mother 9

Another resource, which was discussed by mother 10, was television. A television show was found to be useful as she watched a programme whilst she was pregnant which showed the life of a child with the condition from which her child was going to suffer. She found this helpful as it allowed her to see that life was possible and the condition was manageable. She described the girl in the show with the condition as an “inspiration”.

Mothers 5 and 6 talked about having a doctor within the family. This was helpful as they were able to explain confusing pieces of information which they came across and discuss research. This was found to be very useful for both of them. This kind of support was not available for other interviewees.

Different parents found different forms of and amounts of information helpful and had different levels of education. Mother 11 felt if she had been handed a few small leaflets to go home with then she would not have searched the Internet and found information that she found frightening to read. Conversely, some parents wanted to know everything and studied the research, whereas others were much happier to know very little about the condition. Mother 5 liked looking into stem cell research and could understand most of it as she had studied physiology at university. Whereas, mother 7 felt that studying the condition would just cause unnecessary worry. With the advances in medicine, parents described knowing more about the conditions than their doctors. In particular, mother 3 liked to know everything about the condition. She liked to read all of the research and ensure she knew what was happening.

“I think I know everything that there is to know about it at the moment”- Mother 3

“If I had been given 3 leaflets to read I wouldn’t even have switched on my computer”-  
Mother 11

### Where the parents found support

“Where do we go, where do we turn, who do we look to?”- Mother 9

Some parents were better at finding out appropriate information and accessing support than others. Parents reported help and support being provided from many different areas.

Associations were reported as being a good place to find out information and ask for help. They provided information packs, contacts and other useful resources, which the healthcare team may not know about or have access to. Conferences and new research were available through the associations, which allowed parents to learn about new developments in their child’s condition.

The specific associations were found to be “informative” (mother 4), “brilliant...changed my outlook on everything” (mother 9). It gave parents a chance to network with other families. Mother 4 felt the association was beneficial as it meant her children affected by the condition could meet others who faced similar difficulties in life and it could allow them to feel normal. Mother 6 was part of the association on Facebook and it allowed her to speak to people affected from all across the world as her child had a rare diagnosis. Even mother 10 who did not think the association would be helpful found it a great support network to have available to her.

“I would have kind of scoffed at that kind of thing before and thought oh some people get their kicks out of being part of these big charities and it’s not... I was wrong and they are really helpful so I would recommend it.”- Mother 10

Mother 11 felt the association was beneficial in some respects. They sent her a “red baby book”, which was specific to her child’s syndrome. However, she criticised the way in which she had to enrol as a member. After getting the diagnosis, she then had to prove the diagnosis before gaining access to the site. They asked her to “send a photograph and explain how she was diagnosed”. If she had not taken the diagnosis well, she did not think she would have managed.

As mentioned previously, Facebook groups were discussed by a number of families as a place to gain information and build relationships with others going through similar

experiences while remaining anonymous. Mother 8 felt it was helpful to chat to people through Facebook as it meant she did not have to see other children who were older and affected by the condition but she could talk to the parents to gain advice. Mothers 9 and 11 felt the support provided through Facebook was more personal, from people who were in a similar position.

"You just get real answers back you know as appose to medical answers that are great but it's a real life situation that your living ... if child 11 is playing up or anything, you can just put anything on and somebody will comment on it. It's right through; well it's all over the world that they're coming from so yeah I find that very helpful. I would direct anybody there before I directed them to the foundations page."- Mother 11

Non-specific associations for families with learning disabilities were also seen as being useful, such as "Contact a Family", "Parent to Parent" and "the DDCA" (Dundee Disabled Children's Association). These groups were also able to provide signposts to courses, associations, welcoming nurseries and an opportunity to get to know other people in the same position. Mother 6 described Parent to Parent as being "a great help". Mother 5 liked being able to talk to others who also have children with additional needs and felt that it made no difference what the diagnosis was and whether it was genetic. In contrast to this, mother and father 9 felt that once you had a diagnosis the non-specific organisations were no longer of benefit.

"That's only giving you an understanding of yeah we've both got children with a severe condition but when you've just got a diagnosis, you want to know what the future holds and if they've got a different rare genetic condition then they can't answer your questions that you've got."- Mother 9

Mothers 2, 5 and 7 discussed feeling it was not necessary to be involved in an association. Mother 2 wanted to live a normal life without having to think too much about the condition. Mother 5 did not want to be involved with the association as she felt it was "depressing" and she was happy in her "bubble". Mother 7 felt it was too soon to be involved with the association as her child was so young and not affected significantly enough in her opinion.

“We kind of jolly along quite happily. And the information I’ve got, I still want to just get on and live as normal a life as possible, so I don’t feel the need to use this extra thing to make child 2 stand out”- Mother 2

In comparison, mother 11’s partner did not want to get involved in meeting other families affected by the syndrome, as he wanted to keep his private life private. Also, the meetings were normally in England. However, she felt she would benefit from having the extra support available to her and being able to discuss everyday life with others who face similar problems.

The healthcare team were seen as a good support for families. The “Team Around the Child meetings” in particular were seen as very helpful for mothers 9 and 11 as they provided the family with understanding of who was who. It gave them an opportunity to highlight any area of the child’s life that might need extra care and support. Form filling was seen as an issue for mother 7 and 11 who were provided with help by one of the healthcare team. Mother 7 was also given phone numbers to contact if she had any problems. Mother 11 was provided with emergency financial support when her daughter was admitted to Yorkhill. The paperwork was filled in for her, which she appreciated at such a stressful time.

Mother 5 felt it took them “18 months” to feel they knew who the team was and what their son’s needs were. She felt this was a long time and has now become involved in a group to help the NHS manage parents in a more “proactive” manner.

The provision of sign posting by the healthcare team was an area of management that almost all of the parents felt needed to be improved. Mother 2 felt that she received all of the support she required but only because she asked for it. Mother 3 felt alone and needed some more support and follow up from somewhere but it was not offered. Some parents required signposts to the association and others found them by searching online. Mother 1 wanted to be directed to a support group or association but had not found one at the time of interview. She was concerned at the lack of support she was getting in that respect. Mother 4 felt signposting to support was provided haphazardly by the healthcare team and it could perhaps be provided in a more logical manner to everyone.

Different parents reported different access to support based on their geography, wealth, how proactive they were and who they knew. Some support was provided in England that was not available in Scotland and some conferences were very expensive. Parents discussed the need to fight for support. Mothers 3 and 4 reported wanting to attend conferences with the association but being unable to due to the distance or expense.

Mother 7 was the only parent who felt she was offered more help than she required. She felt she did not need the help at the moment but knows where to go when necessary. She felt it benefited her to know who to contact when she felt it was necessary.

"I have been given numbers of various people I could contact or various websites to go onto but I've never done it because I don't think it's particularly serious at the moment"- Mother 7

### Education and schooling

Certain schools and nurseries were described as being very supportive for the family. Small schools were mentioned as being very good for a child with a disability by mother 5 who felt they were able to get to know the child and support them as appropriate and they were easier to mobilise around. She mentioned choosing a smaller school out of the catchment area for these reasons. Whereas mother 4 felt that at the local small nursery she felt judged by the other mothers. Through Parent-to-Parent she was put in touch with a nursery where all the children had additional needs. She felt this advice had been very helpful. Mother 8 felt that nurseries where all the children had additional needs were more appropriate.

"Through Parent to Parent I was invited along to this playgroup where all the families who went had a child affected in some way. And suddenly you weren't sort of judged, you had a sort of common factor"- Mother 4

Some parents reported having difficulty explaining the child's condition to the school or nursery. The association provided mothers 2 and 3 with information packs to give to the school so that they could understand the conditions that their children suffer from. Mother 2 described teaching techniques being available in the hand-outs in order to help the teachers. She felt it would be impossible to manage if this resource was not available. She

also felt that the mainstream school was very supportive and put a great deal of effort into ensuring her child's needs were managed appropriately. Mother 3 felt similarly in relation to the schools understanding except in relation to her child's Physical Education class, as they did not appreciate her lack of ability in the class.

"The only trouble we have at school is the low muscle tone. It's getting that through to them. It's not that she doesn't want to participate fully in all the physical activities. It's that she can't because she gets so tired so easily and she's not got the stamina" - Mother 3

Some of the children in the study went to mainstream school and others attended schools specific to children with additional needs. Mother 4 discussed her difficulties in considering which school she should send one of her sons to. The head teacher of her children's school told her that a deaf school might be more appropriate for her younger child as he had communication problems. When she visited a potential school she felt it was inappropriate as her child's communication problems were not due to deafness or autism, which appeared to be the main groups within the school. She therefore decided that mainstream school was the best option for him "in the absence of knowing anything better".

Mothers 2 and 3 discussed the transition into secondary school being a particularly nerve racking time. Mother 3 felt the school doctor helped a great deal, as there was correspondence between the school and the doctor before starting school to explain the child's needs. This made her feel more reassured that her child's condition would be understood in the education setting.

### Respite care

Respite care is essential for some of these parents. Respite provides the family with time to themselves. Mother 6 accessed respite care and said it let her "go off and do what a normal 30 year old would be doing". Mother 3 however felt the way in which she had to access respite care was not acceptable. It took her 3 years to get an assessment from social work. She felt that she should have been in touch with respite care earlier so that when she really needed it; it would have been automatically available to her.



“It just took them forever and I joked on the phone that if I was beating her up or if I was an alcoholic I would have had services quicker and I would have done. Had services quicker not beaten her up. But yeah it was a real struggle to get support”- Mother 3

Parents reported childcare being a problem as child-minders may not be happy looking after a child with additional needs or children with NG tubes that need to be maintained. Mother 3 had to hire a nanny as childcare was too difficult to find. This caused financial strain. Mother 6 was able to find a child minder who “was able to deal with disabled kids” through Parent to Parent.

### Family and Friend as support

The parents confessed to not knowing or understanding a great deal when they were given the diagnosis or when they were awaiting a diagnosis. Family members were described as being interested in the diagnosis for many reasons and the parents were the ones to provide them with information. Mother 8 explained that she was constantly asked questions to which she did not know the answer. She felt she could not give them counselling as she was also trying to deal with the diagnosis. Mothers 5 and 7 told their parents not to use the Internet and found relevant information for them that they felt would not scare them. None of the parents were offered advice on how to explain the diagnosis to their family members at the time of diagnosis.

Mother 11 felt she could benefit from advice on techniques that can be used to explain the condition to her other children as they worry whenever they go to the hospital. Child 1’s sibling, who was not affected by the condition, was starting to understand that she was different. Mother 1 voiced concerns that she did not know how to explain it to her. She felt she also needed some advice.

Child 3’s older sibling joined young carers. They were able to help explain the condition to him and provide the support that he needed. Mother 3 accessed this support without direction from the healthcare team. Mother 5 was able to access a course, which she had been directed to through Contact a Family. It was based on how to educate and support a

child with additional needs and the siblings. She found this very beneficial and described how she explained her son's condition to both of her children.

"I've talked to him about how we're made with instructions and some of his instructions have been written in a slightly different way to other people... I got down some wall paper and we drew round him and we said "what do you think you want to write and what do you think about your body".... we did that through with his sister as well just to try and explain and she is only 3, it's just setting the seeds." - Mother 5

The sibling's reactions to the diagnosis were discussed by some of the families. Mother 1 thought her other daughter felt left out as a result of the number of hospital appointments they have to go to. Mother 4 talked about giving her other children extra attention so that they did not feel left out. Mother 3 described her older son being bullied as a result of having a sibling with a learning disability. She thought her son felt held back as a result of his sister and she agreed that he had put up with a lot over the years. She voiced concern that in the future they may not be as close as she would hope. Mother 5 talked about her youngest getting up in the middle of the night and developing certain behaviours, which she felt were a result of her older brother having additional needs and requiring extra attention. As she had done a sibling workshop she felt she understood how best to deal with these behaviours.

There were various reactions to the diagnosis from different family members described. Grandparents were mentioned a great deal. Some of whom provided support and were very involved in the child's life. For example, mother 5 described her parents taking the children when she first got the diagnosis so that the children would not see her upset, which she was grateful for. Other grandparents were less accepting, less understanding and may not want to be involved. Father 9's dad was said to not have taken the diagnosis well. He had a similar grief reaction to that described by the parents and as a result did not get involved in the child's life. Mother 11 also felt her parents took the diagnosis badly.

Other reactions described were Mothers 2 and 3s' parents who were thought to be in denial. Mother 2's parents thought the condition was something that could be "grown out of". Mother 3's parents thought "better parenting" could fix all of the problems. Mother 10 described her mother obsessing over her child's disabilities and praying that she would get

better. Mother 8 explained why she felt her parents did not take the diagnosis well. She hypothesized that grandparents want to spoil their grandchildren, thus as they could not do that with her child, they felt disappointed.

"He doesn't play with toys; he is very much in his own little world...And everyone is going what will I get child 8 for his Christmas, what will I get him for his birthday and I've got to the stage where I, you know... I just don't want to sit in a house full of toys that he doesn't play with as well because that's just as hard"- Mother 8

Family members acted as an essential support for most of the parents. They helped in the care of the child. Some of the parents talked about their parents getting older and frailer and their worries relating to not having that support available any longer. Mother 6 was concerned about when her mum was no longer around to help. Mother 4 wanted to have four children, as she felt this would mean her two affected children could be more easily cared for by their two unaffected siblings. Mother 2 discussed getting older and concern as to who will look after her daughter once the older family members are gone.

"We'll be getting older and who'll be able to help her when we're not basically you know further down the line." - Mother 2

## How the genetic diagnosis affects the family

### The impact of the conditions being genetic

As the conditions were genetic, other family members were described as having concerns for their own children and what it meant for them. For example, mother 1's sister was pregnant at the time of diagnosis, which caused her some concern. The genetic diagnosis also led to blame being passed as to where the faulty gene may have come from. Mother 4 knew the gene had come from her partner. They had explained this to his family and how it related to the family history of cardiac events. Mother 4 described the family as treating the diagnosis like a "dirty secret" and wanting to "push it under the carpet".

"When we got the diagnosis for our immediate family, we immediately told my mother in law ... I don't know maybe she was just in denial or what but she was very resistant to what we were suggesting." - Mother 4

Mother 5 seemed to blame the father of her son, from whom she was separated, for the genetic condition. She wanted to be able to explain to her son where the gene had come from. Mother 5 also explained how it felt when she realised her son's condition was genetic and she did not know whether she was a carrier. At the time, she had a daughter with a new husband and was therefore concerned her daughter could also be affected.

"Getting results about me not being affected and therefore our daughter was really it helped... because my daughter is his biological daughter, that was quite hard. But he was trying to support me but I could see he was struggling as well" - Mother 5

### Family relationships

Parents described different reactions to the diagnosis within their relationships. Many of the mothers were no longer with the father of the child. Mother 6 was divorced and described her ex-husband not coping with the condition, which "led to our marriage breaking down". Her ex-husband had not seen his son in 2 years at the time of interview. Mother 3 was advised to terminate her pregnancy, which she did not want to do. Her husband felt they

should terminate. This argument “led to a lot of problems” and divorce. Mother 1 described her relationship ending as a result of having a child suffering from a genetic condition. This resulted in a need for marriage counselling.

“Went through a bit of bickering... we went through a phase of that, we didn’t speak we just didn’t manage... quite a bit of pressure on our relationship. Cause we analysed ourselves... We went to marriage counselling. We were falling apart” - Mother 1

Mother 2 suggested the diagnosis might have made their relationship stronger as they knew it was something they had to get on with as a couple. However she described not being able to talk about how she felt about the diagnosis with her husband. They would discuss their child’s problems but would not talk about how they were coping with it. She developed postnatal depression.

“We’re not kind of the sort of couple that’ll sit and pull it to bits and think about it. We kind of did the opposite; we went opposite direction with it... It was something we had to get on with together” - Mother 2

### Friendships

Friends can play a big role in ensuring families feel normal. Mothers 7, 10 and 11 felt that nothing had changed with their friends since having a child with additional needs. Some parents said that their friendship group had changed as a result of having a child with additional needs. Their friends became the other parents who also had children with additional needs. Mother and father 9 felt their old friends pitied them and they did not like that. They felt it was easier to speak to other parents who were in the same position as them. Mother 8 said she thought her old friends found it difficult to speak to her, as they did not know what to say when they found out. She described being out with her old friends more recently and not having anything to say to them as so much had changed. She felt she had lost a great deal of confidence as a result.

Mother 5 discussed the “Welcome to Holland” poem, which she felt describes perfectly how families feel when they have a child with a learning disability. They are in Holland and all of their friends who do not have children with disabilities are in Italy.

“All your friends are in Italy and they are all having a great time and, you know all your friends that don’t have additional needs children, and you go on there and actually you are in this other little world and Holland is a beautiful place but it’s not Italy and there are times when you can feel a bit isolated” - Mother 5

Some parents wanted to explain the diagnosis to all of their friends. Mother 4 felt it was easier to be “open and honest about it rather than having people whispering “Oh, why isn’t he talking yet””. In contrast to this, mother 1 felt very differently and did not want to discuss it or tell people about it. Mother 5 felt similarly, she did not want her son to be labelled, and so she let most people think her son had cerebral palsy unless they were close friends. Mothers 6 and 9 felt it was pointless trying to explain, as nobody understood what it meant anyway.

Mother 1 described being approached by a stranger in the supermarket and being asked what type of cancer her child had. She would get asked when she was in town what was wrong with her child. She said this “would kind of bring back a lot of what had happened”.

### Family planning

All of the parents agreed that they understood the risk of recurrence in future pregnancies. They had been given information from the healthcare team. Mother 8 had been given a basic diagram of a sperm and an egg and a picture of the chromosomes that she found very helpful.

There were many different opinions on family planning. A number of parents had decided not to have any more children. Some parents decided this because they simply did not want anymore. Mother 7 thought she would never be a mother and was more than happy with one child. Other mothers did not want to have further children with similar problems. Mothers 1 and 3 knew the risk was small and wanted further children but they had decided

not to go ahead in case there were further problems. Mother 2 appeared to have similar reservations after being told that the risk was very small. They opted to wait a few years and then went ahead and had a further child. Mother 10 did not want to repeat the bad experiences which she had during pregnancy and felt she could not have another child with a disability as she would have less time to give to her other children.

“Just from what I went through, I don’t know if I could risk having another child with something wrong...I wouldn’t want to have another child with additional needs and then not have I suppose the same attention to give to her and to give to my little boy. So, I don’t think we’ll be having anymore” Mother 10

In relation to testing during pregnancy, many opinions were raised on this subject also. Mother and father 9 thought they would ask for testing in their second pregnancy but during the pregnancy they decided there was no point, as it would not change what they would do. They were offered testing for their newborn child but at the time they knew that their new baby did not have the same condition so again declined testing. Mother 6 said she would consider having another child and did not think she would want testing in pregnancy either as she felt “there is no way that I could get rid of it”. Mother 2 opted to have an amniocentesis with her next child “just to check”. She found this a “huge relief”.

Few parents discussed termination of pregnancy. Mother 3 described getting the diagnosis as “what life gives you. It’s a bit of a pain but you just deal with it as it comes”. She was offered a termination but did not want it.

Mother 4 decided to have prenatal genetic diagnosis (PGD) as the father of her children was affected. She felt her fourth child was her “little miracle”. When asked about this further, she said she did not give PGD a great deal of consideration at the time. She knew she wanted another and their time was running out so they went for it. However, after having the child, they stopped and thought about the morals behind the test. They did not regret it and she felt positive that the genetic condition could be cut out in one generation. However, the embryos were made and then discarded which she felt morally unsure about. She opted to have further children, as she wanted extra support for her affected sons when they are older. She also felt she was providing them with an option for the future if either of them wanted children. They could use their “mum as a role model”.

"The creation of all these embryos.... it only just hit me much later on "did we really do that? Should we not have given that more thought?" Just as well we didn't at the time. I suppose as the boys get older and you see their characters grow more, you know just how special and wonderful they are, and you kind of god you know, there were potentially 7 others" - Mother 4

Mother 1 was the only parent to discuss siblings having children. Her older daughter was not going to be tested for the gene mutation until she was 14. The parents wanted her to be tested earlier as she could be sexually active by that stage. She also discussed her child, who suffered from the genetic condition, having children and felt it was not something they needed to worry about yet. She felt that as "there's lots of research being done", more management options could become available.



## Discussion

After eleven interviews the main themes from the interviews saturated and complemented previous research. The different themes, which have been described above, presented a number of interesting aspects of parents' reactions to receiving a genetic diagnosis for their child. The relatively high turn out for interviews suggest that parents are keen to have their say on how they were managed. These experiences highlighted a need for a management plan to be put in place for the families who are affected by a genetic diagnosis. This discussion will hopefully make readers aware of the importance of taking future parents' needs into account when managing them.

### The diagnosis

Two articles by Lewis et al and Rosenthal et al examined how parents of children with different syndromic features cope without a diagnosis (14, 21). They found parents suffer from a significant grief reaction, feelings of being out of control, anxiety relating to and an inability to come to terms with the unknown. Our research complements this as parents described similar feelings prior to obtaining a diagnosis. Mother 9 felt not having a diagnosis was like being in "limbo". The unknown was associated with feelings of guilt relating to the cause of the condition, for many parents, and whether they could be to blame. One example of this was mother 7's concern that her epilepsy could have caused her son's condition. In terms of feeling unable to come to terms with the unknown, mother 3 described how her worry relating to her child's future affected the development of their relationship.

Parents in this study and in a study on children with Fragile X syndrome described the different ways in which a diagnosis was given (10). There did not appear to be a set protocol on how to give a diagnosis and the information and follow up that is required, despite it having a significant impact on parents' experiences. Paediatricians, in research by Turner and Sloper, described the importance of getting a diagnosis early in order to improve management (23). Mother 11 received her child's diagnosis before birth, which enabled her to access information and feel more prepared. Both paediatricians and parents of children with Down's syndrome have highlighted the need for a diagnosis to be given in the right

setting and with good communication skills, information and follow up. Hasnat and Graves highlighted the impact of communication skills on parental satisfaction (25). Mother 5 described being told in a rushed setting without appropriate communication skills and follow up that her child had cerebral palsy. She was upset about the way she had been managed on this occasion. Her experience of receiving the genetic diagnosis a few years later was significantly more positive. On that occasion, she was given the diagnosis by her paediatric consultant who knew her and her family well, with time to discuss what it meant and with her partner present. This highlights what was found in the previous research, as the manner and the timing of the diagnosis significantly affected her experience (10,23,25).

Receiving a diagnosis was seen as beneficial by all of the parents involved in this study. Prior to the DDD study commencing, parents' reasons for wanting a diagnosis were researched. Gaining a label and answers to questions about the child's condition were thought to be important reasons to search for a diagnosis (13). Parents in our study agreed that having a diagnosis does ease the curiosity around the condition, which their child suffers from. For example, mother 3 said when she got the diagnosis she "joined the support groups, got a book on it, look(ed) at research papers". This allowed her to learn about the aetiology and prognosis of the condition and allowed her to plan for the future.

Being able to provide an explanation of their child's condition and the risk of recurrence to friends and family was thought to be an important reason to gain a diagnosis in previous research (14, 15). Some parents within this study agreed. However, a number of them mentioned that the rarity of the conditions, which were involved in this study, meant the diagnosis was not always helpful. For example, mother 9 described the diagnosis as "a name floating".

One of the main benefits to diagnosis mentioned by the parents was increased access to support including financial, informative and groups. Some parents we interviewed mentioned not realising support was available until they had a diagnosis. In a study by Lenhard et al, it was found that parents with a diagnosis of Down's syndrome were more likely to access support groups than parents of children who did not have a diagnosis (15). Thus, having a diagnosis appears to open doors to support. This may suggest that support is

more widely available for specific conditions or it may be that parents without a diagnosis are less likely to be sign posted to support.

Also in Lewis et al research, a parent described knowing the diagnosis as being helpful for her everyday life and getting appropriate housing and education organised (14). Mother 10 described how she was able to access information on how to make “home-made gadgets” which could help her child with simple things such as using light switches. She was able to access this through the specific support group which she would not have had access to without a diagnosis.

Parents also described how a diagnosis could have a negative impact. Mother 8 found the prognosis of her child’s condition difficult, as she had hoped her son would get better. This feeling was also described by parents of children with Ataxia Telangiectasia who felt the diagnosis made them lose hope (17).

Other parents simply found the diagnosis unhelpful. Lewis et al interviewed a mother who described the diagnosis as being like a foreign language (14). Many of the parents within our interviews were also initially frustrated about not understanding what the diagnosis meant and described hearing “the locus of blah blah blah” and “micro RA something, some complicated thing with all the genes” from their doctors.

Grief reactions have been discussed in previous research in relation to a genetic diagnosis (19, 26, 27). The parents within our interviews described grieving for various reasons including not wanting their child to suffer or miss out on normal everyday activities and feeling that they had lost the child that they initially thought they had.

It has been hypothesised by Ciske et al that parents of children with developmental delay from higher socioeconomic groups may not cope as well as parents from lower socioeconomic groups (52). In addition to this, it was found in research Hoare et al. that the severity of the child’s condition could have a negative impact on the parent’s mental health and Griffith et al. that the rarity of the diagnosis increases parental anxiety and depression (31, 33). Mother 6, who appeared to be denying the significance of her child’s problems, was in socioeconomic group 8 (10 being the highest), her child’s condition was severe and there were less than 1 in 1,000,000 sufferers world wide. Therefore, from previous research,

mother 8 would be more likely than other parents within the study to develop mental health issues.

Some of the parents openly discussed the affect of the diagnosis on their mental state. Mothers 2, 3 and 4 described their experience of postnatal depression and mother 3 also described the father of her son becoming suicidal and requiring inpatient psychiatric treatment. Mothers 2, 3 and 4 were in socioeconomic groups 6-8 (10 being the highest), thus being near the higher end of the socioeconomic spectrum. This helps to reinforce this hypothesis made by Ciske et al (52). However, their children's conditions were all rated mild-moderate which goes against the theory made by Hoare et al (31). In contrast to this and supporting the theory made by Hoare et al, mother 7 remained positive throughout interview about her son's condition and did not discuss any problems with her mental health. She felt her positivity related to his delay being motor only.

Many of the parents, in the interviews, described a 'bubble' that they lived in. The term 'bubble' was used to describe feeling both restricted from normal everyday life by their situation (mother 8) and also how they tried to protect themselves from planning for the future and becoming emotionally overloaded (mothers 1, 5, 8, 11). In previous research by Coyne and Canam, parental denial when a child was diagnosed with Cystic Fibrosis or other chronic condition was initially seen as an effective coping mechanism (29, 30). It allowed parents time to gain an understanding of their situation and develop useful long-term strategies (29, 30). Interestingly, mothers 9 and 10 described being in a 'bubble' initially, before attending a conference. They both described the conference as being a turning point in which they realised that other people managed their children's problems and therefore they could as well by using alternative coping mechanisms.

Parents of children with Ataxia Telangiectasia turned grief into feeling lucky to have a child who was different (17). Three of the parents in our interviews described similar coping techniques. Mother 7 felt 'lucky' to have a child, mother 6 felt 'special' having a child with a condition that is very rare and mother 4 felt 'proud' to be able to watch her children overcome their obstacles. Mothers 7 and 9 both felt the comparison to other families made them realise that their situation was manageable. This positive thinking towards the diagnosis appeared to be a common coping mechanism.

From this, it appears that having an understanding of the parents main concerns and problems in relation to a genetic diagnosis could allow for better management of these families. This could significantly improve their experiences and ensure their needs are met.

### Hospital management

Parents reported having a large number of hospital appointments to attend, with a number of different specialities, concentrating on different areas of their child's health. This was time consuming, stressful and confusing trying to understand who was who and why they had to see another new healthcare professional. Lewis et al described parents having to repeat their story to different healthcare professionals, which parents could find distressing (14). This was an opinion that was shared by a few of our parents. In particular, mother 1 who decided to stop attending hospital appointments all together as a result of this. She explained: "I'm not going to sit and explain child 1's whole life all over again". Additionally, she felt the number of appointments she had caused problems living her everyday life. Parents within this study reported changing job, loss of income, loss of social life and siblings feeling left out as a result of the number of appointments.

In order to try and overcome some of the above management issues, the parents identified solutions. For example, many parents felt that having a single key worker could be beneficial to act as one main point of contact. The need for a key worker has been reported in previous research involving parents and paediatricians (14, 16, 23). Key workers would co-ordinate care, develop a care plan, provide valuable advice and signposting to additional support and information. Mother 11 felt she had this service from her "Parent to Parent" support worker and appeared to really appreciate the help and support given to her.

Parents discussed the time it took to become comfortable within the healthcare team. They found that they did not immediately have a good support network around them. Many of the parents described initially feeling like they were in another world. As they became more knowledgeable about the condition, the management and the support available, parents gradually became more comfortable. If parents had a key worker the process of being introduced into this new world may be made easier.

Mother 8 attended a specialist clinic in which all of the healthcare professionals, that needed to be seen, could be seen within one day at the hospital. Additionally, they all understood what the diagnosis meant. Mother 4 had heard that this type of clinic was available for her children's condition in other locations in the UK. She felt this would be very beneficial for her.

Due to the rarity of the conditions involved in the study, some of the parents felt their doctors knew very little about the conditions that their children were suffering from. Mothers 6, 8 and 11 had similar experiences of doctors looking up the diagnosis while they were in the room or asking them directly what the condition means, as they had not heard of it. Paediatricians often play a role in counselling and improving the parents understanding. Despite this, Skirton also described parents feeling the healthcare team lacked knowledge in genetics (16). Many other studies have reported the benefits of having a specialist in genetic counselling supplying information to families, as they have been trained on how to provide the relevant information in an appropriate way (34, 35, 36, 41, 42). Yet despite this research, studies have found that only 7.4% of 230 children with genetic metabolic disorder and only a quarter of 18 children with Neurofibromatosis 1 were referred onto a genetic specialist (19, 36).

Most parents reported that their children were being well looked after clinically. However, they felt healthcare professionals did not always take into account the emotional impact of the condition and the need for extra support. Parental satisfaction with the health care provision has been found to directly relate to the empathy and communication (24). A lack of or delay in follow up and a lack of sign posting to the support available were other main complaints. This was also described by Albon who identified that children diagnosed with Neurofibromatosis 1 were not routinely given follow up, despite the parents feeling it was needed (19).

Many of the parents would have liked more contact, a phone call after the diagnosis or an appointment a week later, to discuss the diagnosis once the initial shock had sunk in. They wanted to be able to talk to someone who had time to listen to their concerns and feelings about the condition and who could explain what it means for them and their family. This did not need to be a doctor. These results suggest a need for paediatric genetic counsellors and

key support workers within Tayside. Around the time of diagnosis, families will be having a large number of medical appointments. Interventions at these times could ensure problems were picked up early and support services could be put in place.

### Access to support and information

The experiences of parents within our study identified a need for further consideration of the information that is given to parents at the time of diagnosis. As the conditions the children suffered from were rare, the parents did not automatically understand the meaning of the diagnosis. They described how difficult it was to take in information at the time of diagnosis and they became quickly confused. This identifies a need to provide appropriate information on the diagnosis and what it means for the family, which can be re-reviewed.

The parents in this study described being given a range of information from none to a pile of papers to take home. Neither of the extremes benefited the parents in their opinion. This finding was documented in previous studies, which identified that knowing how much parents want to know about a diagnosis and avoiding overloading them with information is important (20, 25). A few studies found that information in written letters after the consultation and leaflets on a diagnosis can be beneficial (16, 46-47).

Barr and Millar researched the information which parents were given when they were initially referred to the genetics services and they found that parents were not given enough information at this stage either. They hypothesised that this was because the doctor referring them lacked knowledge on what would happen when being reviewed by a geneticist (16).

Petrucelli et al identified that 77% of 26 parents, who had been given a diagnosis of a foetal sex chromosome abnormality in pregnancy, felt they had a poor understanding of their child's expected diagnosis (45). Other studies have documented that parents' often have a poor understanding of basic facts on a diagnosis for example risks of recurrence. Parents were prone to underestimate their risk in both ataxia telangiectasia and Cystic Fibrosis (17,

27, 52). In contrast to this, all of the parents within this study felt they had a good understanding of the risk of recurrence. They were not asked to explain their risk in full to prove their understanding, however this may show that there has been a change in the information provided to families as a result of the previous research.

The Internet was used as a main source of information for the parents. The harm that can be done when searching the Internet without having direction to reliable sources has been documented (14, 19, 34). The information gained can be overwhelming and inaccurate. Many of the parents had negative experiences of searching online without direction and described finding worst-case scenarios, especially on American websites. Additionally, some parents found it difficult to tell whether information was relevant for them, as many of the syndromes can vary in the way they present and how the children are affected. Phrases such as "demented" were used to describe how it felt to read through research. Waitzkin found that patients who were given adequate information by their doctor were less likely to search online (51). This could be very difficult to get right as parents within our study wanted to have a varying amount of information.

In contrast to the negative experiences parents had on the Internet, Facebook was highly recommended by some of the parents. They explained that Facebook provided a safe space to discuss their child's problems with families affected from all around the world. This was particularly beneficial for mother 6 whose child suffered from a condition with a prevalence of less than 1 in a million. Mothers 6, 9 and 11 described being able speak to other parents who were experiencing similar problems and gain advice and support from people who truly understand their situation. Parents felt this type of information was invaluable.

Another source of information, which mother 10 described, was a television show. She received a diagnosis for her child during pregnancy and felt seeing a child with the condition was "an inspiration". She watched the child on the TV show manage everyday problems, which made her feel more positive about the future. Parents of children with sex chromosome abnormalities were shown pictures and given information on what to expect which they also found beneficial during pregnancy (34).

Parents reported associations as being good places to find information and ask for help. Parents received information packs, baby books specific to the child's condition, contacts



and other helpful resources, which the healthcare team may not know about or have access to. Conferences were also available through associations, which allowed parents to learn about new research on their child's condition and meet other families affected. Previous research looking into the benefits of parent led support groups identified parents developed new skills and got a sense of power, confidence and belonging from attending groups (43, 44). Additionally, couples appreciated meeting parents of children with similar conditions as they grew a better understanding of what to expect (34).

Non-specific associations for children with learning disabilities were also seen as being useful. "Contact a family", "Parent to Parent" and the "DDCA" were all mentioned and seen as being an invaluable resource. These groups were able to provide signposts to courses, specific associations, nurseries that were appropriate for children with additional needs and an opportunity to get to know other people in similar situations. This shows that support is available for families before they have a diagnosis for their child.

Some parents did not want to get involved in the associations because they wanted to keep themselves to themselves or because they felt it was unnecessary. Mother 10 admitted to making presumptions about associations and the type of people who would be involved and this had put them off initially. She later found that having an association available was helpful.

Financial support was available for the children and their carers. A number of the parents described a delay in claiming the financial support available to them, as they did not know they were eligible. Some had to "fight" for the benefits they were eligible for, whereas others were given advice from different healthcare professionals or support workers on how to access them and complete the forms. This advice appeared to be given inconsistently.

Skirton has previously documented a lack of referral and signposting to this type of support (16). A lack of signposting to information and support available was also highlighted as a problem for many of the parents within this study. Mother 4 suggested that a checklist could be provided to parents to ensure they were accessing all of the help that is available to them. It could inform parents on how to access financial support, childcare and respite,

organisations, and reliable information. This could result in a more consolidated approach to providing sign posting, thus ensuring that none of the parents miss valuable advice.

From this and previous research, it has been found that information which parents are given surrounding genetic diagnosis is often inadequate and this can leave parents with a poor understanding of the genetic diagnosis. There are many ways in which parents can become informed and parents differ in how much they want to know and what forms of information will be of most benefit. Parents' previous experiences suggest that this should be taken into account when providing information. Advice on where to access further information and support needs to be provided. The parents felt the main benefits of having a diagnosis was that a better understanding of their child's diagnosis could be gained and better access to support was available. Thus, inadequate information and signposting could make having a diagnosis pointless in this respect.

### The wider family

The family network, which the child and parents have around them, plays an important supportive role. Many of the parents discussed getting emotional support and help with caring from different family members. Mother 4 decided to have IVF so that two of her children with additional needs would have two siblings who did not have the genetic condition. She felt having a larger family would help to ensure that her children with the genetic condition were looked after when they were older.

In contrast to this, having a genetic condition causing a disability within the family can cause increased strain on relationships. Parents within this study described family members grief reactions and their need for support or counselling at the time of diagnosis. In particular, grandparents were described as having difficulty accepting the diagnoses. Mother 8 felt this was because they wanted to spoil their grandchildren and were not be able to do that easily.

Furthermore, due to the diagnoses being complicated for the parents to understand, explaining the diagnosis to other family members was described as being very difficult and

caused initial stress. It was mentioned that having an information sheet for family members could prevent this becoming a problem. One mother's reflection on her experiences of having a child with Adrenoleukodystrophy reiterated the difficulties parents could have providing information to family members while trying to come to terms with the diagnosis (18).

Some parents were given advice on how to support their other children and describe the condition to them. Mother 5 was sent on a course on how to manage the siblings of a child with developmental delay. Young carers provided support to child 3's sibling, who was bullied at school as a result of having a sibling with developmental delay. These were resources that other families may have benefited from. Mothers 3 and 6 echoed Lewis et al who described the restrictions that families can have on activities that they are able to do with a child with developmental delay. This was reported to cause resentment from the siblings (14).

Due to the conditions being genetic, family members were interested in how the condition had been inherited and what the risk was for other family members. In two separate papers discussing Muscular Dystrophy and Cystic Fibrosis, guilt and blame were mentioned in relation to identifying the causative gene mutation (27, 54). Mother 4 felt her family did not want to know where the gene mutation had come from. She felt her family treated the diagnosis like "a dirty secret".

Breakdowns in the parents' relationships were described as being the result of having a child with additional needs causing strain or the genetic nature leading to feelings of guilt or blame. Only mother 2 felt her relationship grew stronger as a result of their child's additional needs. Wei and Yu also found that single parent families were more common if a child had certain disabilities (55). This may relate to different ways mothers and fathers react to a diagnosis. For example, mother and father 9 felt they reacted differently to the diagnosis, while she became very emotional, he felt he had to be strong. Another study identified that mothers are often more anxious than fathers. This was thought to relate to the mother acting as the main carer, attending most of the hospital appointments and acting as main information provider to other family members (28). In addition to this, fathers of children with different genetic syndromes have been found to be less emotional

and more distant (17, 19). Within this study, the fathers were invited to the interviews if they lived with the child. However, only one father attended.

All of the parents felt they received good explanations on the risk of having future children affected by the same condition. Diagrams and simple written information in letters post diagnosis helped understanding.

Some parents decided to have further children and some did not. The reasons mothers 1 and 3 gave to explain their decision not to have further children included concerns about future children being affected by the same condition despite knowing the mutations were *de novo*. Out of 73 parents of children with spinal muscular atrophy the vast majority decided not to have further children and the main reason given was the risk of having further children affected by the condition (39). However, the risk was often much less significant than they assumed (39). This may be a result of not having a true understanding of their risks.

It has been found that stress levels of parents, child behaviour, and social support concerns for the child's future directly affects how the parents feel towards future reproduction and the decisions that they make (36). The age of the couple, condition of the child and number of previous children also affected parent's decisions on termination (56). Within this group of parents, none said they would consider termination for future pregnancies. Mother 3 had testing for her next pregnancy, which provided her with "relief", but she felt she could not terminate a pregnancy.

Mother 4 opted for prenatal genetic diagnosis and felt her daughter was a "miracle". She said she has only considered the ethical aspects relating to this decision more recently and not before making a decision. She voiced her distress at discarding the remaining embryos that she felt were like her sons. In one mother's reflection on her son's diagnosis, she discussed the need to talk about the ethical repercussions of different management options with a specialist in that area. She felt she was given the factual information but not the ethical (18).

### Limitations of study

The interviews discussed parents' retrospective memories of their experiences of having a child diagnosed with a genetic condition. Many of the questions asked were targeted at the time of diagnosis, which was months or years previously. This time lapse may have skewed parent's opinions and there may be some discrepancies. The drawback to using semi-structured interviews was the risk of influencing the parent's opinions in the way the questions were asked. Some of the interviews took place within the Clinical Research Centre and some took place within the parents' homes. This may have lead to bias. The use of a small sample size means that points made cannot be generalized for other parents in the same situation. Parents who were particularly involved in research were more likely to get involved with this project, perhaps leading to biased results. Findings suggest a lot of common feelings between different diagnoses, which suggest that cases can be compared.

### Implications for practice

Many suggestions were made by the parents on how to improve management for future parents who have a child with additional needs diagnosed with a genetic condition.

1. Key worker – A named person who would be available to provide advice, support, signposts to organisations and information.
2. Follow up appointment/ phone call one week after diagnosis – This would provide an opportunity to discuss the diagnosis after the initial shock has eased and reading information on the condition. It would provide an opportunity to ask questions.
3. Specialist clinics – This would enable parents to see the relevant consultants and allied health professionals within one session.
4. Leaflets – To provide parent and family friendly information on what support is available and what they need to know.
5. Closer links with associations – To ensure the healthcare team signpost to the support networks that are available.
6. Checklists for parents – To ensure parents know what support they can access and may benefit from.
7. Psychological support – Available for parents who feel that they require a professional to speak to about how they are coping.
8. Benefit support – To ensure parents receive the financial help they are eligible for.

Lack of resources may make some of these suggestions difficult to put in place as more clinic appointments, staff time and finances would be required. As many of these conditions are rare, specialist clinics may be aimed at a small cohort of people and it may not be viable.

Further development and dispersal of the questionnaires, which have been developed and are included in the appendix, may provide more information on how consultants feel they manage these families within NHS Tayside. Additionally, if any of the above management suggestions were put in place, the questionnaire could be altered to target parents and an audit cycle could begin. The results will be disseminated by presentations, publishing in a peer reviewed journal and parents involved will be given a parent friendly version of the paper.

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Division of Medical Sciences  
Centre of Oncology & Molecular Medicine

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Protocol

1. General Information

Parents' experiences of genetic diagnosis in their child: an exploratory study

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2. Summary

This study aims to find out how parents react to their child being diagnosed with a genetic condition and what their experiences were at the time of diagnosis. The information will be gathered qualitatively in semi-structured interviews and themes will be identified.

3. Background

Whilst doing research, I found that very little information exists in the literature on how parents react to and cope with having a child diagnosed with a genetic condition. Different parents will react differently and will require different amounts of counselling and support. However, at the moment, research on how best to manage and support the parents is limited. If we do not research what these parents needs are and how best to provide them, then the healthcare team will not be able to offer them the service which they deserve.

Genetic diagnosis is becoming much more efficient. In the past 20 years genetic tests have become more accessible and more commonly used in investigating a child who may be suffering from a genetic condition. In the past, genetic conditions were diagnosed on clinical features alone. The completion of the human genome project will allow more genetic testing to become available. Therefore, with our increasing knowledge of genetics and genetic conditions and our increasing ease of diagnosis, it is going to become increasingly more important to know how to manage the parents of children with a genetic condition.

Diagnosis has proven to be beneficial for the prognosis of the child, as it means a management plan can be put in place, and all health care professionals, which may be able to help, can be involved. The diagnosis will affect the parents as the implications of having a child with a genetic condition are huge. Parents will be required to look after their child who may have learning disabilities and other associated health problems.

This may mean they have to attend a large number of hospital appointments, which can make social life and working life difficult. It may also lead to financial troubles. What's more, family planning issues need to be addressed. Understanding this can be very difficult. Parents may not realise how their lives will be affected.

Knowing how to communicate the diagnosis with parents may be difficult. A number of studies have identified the importance of good communication skills in all types of medical practice [1-4]. Communication can improve patient understanding, which has been found to decrease patient anxiety, improve patient satisfaction and increase the chances of the patient following the management plan. This may improve the prognosis of the condition. In addition to this, if patients understand a sufficient amount, then they may not need to look up resources in the media or the internet, which may be confusing, inaccurate or exaggerated.[4]

It has been found that patients often lack a basic understanding of the condition which their child is suffering from and what implications the diagnosis carries with it. For example, it has been found that only 57% of parents, of children who have been diagnosed with cystic fibrosis, understand that their child has a 25% chance of having a child with Cystic Fibrosis, if they reproduced with another carrier[1]. This lack of understanding should not be acceptable. People should know the implications and should be able to make informed decisions. This may be a direct result of bad communication skills or a lack of understanding of the parents' needs.

Parent's lack of understanding may be a direct result of inadequate management. For example, a great deal of information is given at the time of diagnosis, which is a particularly difficult time for the family, and it may be too overwhelming for the family to deal with. Parents may be worrying about immediate health concerns. It may be beneficial to give only the necessary information right away, then have follow up appointments to add additional information on the implications of the diagnosis e.g. family planning[2]. Many parents decide not to have more children as they may assume there is a large risk. These assumptions may be made without obtaining any facts, as they may not access further genetic counselling[2]. Families should perhaps be given further check-up appointments to ensure they have all of the information and advice which they need.

It has been reported that parents' views on the genetic counselling which they receive varies significantly depending on the member of the healthcare team who provides it[2]. For example, neurologists, who may not be trained in genetic counselling, have been found to leave a more negative memory of the experience than qualified genetic counsellors. Neurologists, in this study, were able to provide a great deal of factual information but they did not necessarily know what was best for the parents. They were "perceived as having the poorest understanding of parents' needs".

It has been found that many paediatricians do not know how to deal with the families of children diagnosed with a genetic condition[3]. Paediatricians could play a role in counselling and improving the parents understanding, but it has been found that they

lack the knowledge on genetics and the appropriate communication skills[3]. Doctors who are expected to deal with genetic conditions on a regular basis should be properly trained to deal with the families and their questions. They should understand the genetic diagnosis and should be able to explain it in a manner which the parent will understand. The way this is achieved will vary between parents.

“Additional research is needed to determine whether there are communication styles and approaches that are better suited to counselling parents”[1]. It has been recognised that there is a lack of research and knowledge within this area. However, the research out there remains very limited. Parents may react differently to being told their newborn baby, which they believe has no health concerns, has a genetic condition; compared to parents, who have coped with a child who has been developmentally delayed for years, being told their child has a genetic condition. The latter may feel a sense of relief at being given a diagnosis and an explanation, even if the diagnosis is not going to affect the management of the child. Other parents may react badly, even if the child has a good prognosis, parents may hear a diagnosis and automatically think the worst. The diagnosis can be devastating for parents; therefore, parents should be provided with as much help and advice as health professionals are able to give.

#### 4. Study Design

To find out how parents react to being told their child has a genetic condition and what their memories of the experience are, a qualitative approach will be employed. This approach will be conducted in the form of semi-structured in-depth interviews. This will allow for a wide range of views and perceptions to be identified. Following analysis will allow these views to be conceptualised in a useful way. It is a suitable method for the identification of emerging key issues. The information gathered will be on the parents’ retrospective experiences of genetic diagnosis.

#### 5. Study Aims

This research will help identify what parents’ experiences of genetic diagnosis in their child were, how parents react to their child’s diagnosis and key points which should be addressed when giving parents genetic counselling, advice for the future and managing them in general.

#### 6. Sampling

The participants will have a child who has been diagnosed with a genetic condition which causes a developmental delay and learning disabilities in the Tayside area. All participants will be over 18 and be able to give informed consent.

#### 7. Methods

Interviews will take place on a one-to-one basis with Lisa Bryson in the Clinical Research centre, Ninewells hospital. If, for whatever reason, this is impossible, interviews will be conducted in the individuals’ home with the appropriate safety



precautions being put in place. Interviews will last a maximum of one hour and will be digitally recorded. Qualitative analysis will aim to explore and identify common themes and issues within the data. The data source will be transcripts from the interviews. The "Framework" method of analysis will be used for the interview transcripts. This includes a systematic 5 step process. Both the student and chief investigator will be involved in the analysis.

#### 8. Subject Recruitment

This will take place through the NHS Tayside, Clinical Genetics Department. Eligible patients will be identified through the clinical genetics records. A member of the research team will carry this out. Potential participants will be contacted via letter from myself and informed of the project. If no reply slip has been returned after 3 weeks a further reminder letter will be sent. Further discussion and completion of an informed consent form will take place prior to the interview. Ideally 4-6 participants will be interviewed in the clinical research centre, Ninewells hospital. Confidentiality will be maintained by the anonymisation of participant's details.

Risks for taking part in this study are minimal; however discussing personal experiences of their child's diagnosis may lead to increased anxiety/ distress for participants. Further contact with the Clinical Genetics team will be offered routinely at the end of each interview. In addition it is possible that participants could experience some minor inconvenience in attending an interview. Steps will be taken to ensure that interviews are, as far as possible, held in a venue, and at a time, which is convenient to individual participants.

#### 9. Data collection and record keeping

Transcripts from the recorded interviews form the data set for this project. The recordings of the interviews will not contain patient identification details except for first names. This will later be assigned an identification number. Full transcripts and questionnaires will only be available to Lisa Bryson and the chief investigator. All data will be anonymised and kept in password protected computers. A paper record will be accessible to Lisa Bryson until completion of the project. Quotes, categorised by theme and therefore unidentifiable, will be stored while publication of the results is being sought. Results will not contain any identifiable personal data so all quotes will be devoid of any detail or contextual information that might indicate an individual's identity. Recordings of the interviews, patient identification numbers and full transcripts will all be destroyed following the completion of the study.

#### 10. Analysis

All interviews will be audio-taped and fully transcribed. Transcripts will be analysed using framework analysis. Framework analysis is proposed for several reasons. Primarily, it is particularly suited to conducting applied qualitative research. Applied research, unlike basic or theoretical research, is often required to meet specific informational needs. It provides a greater understanding of the issues addressed and

suggests strategies to affect outcomes. Secondly, framework analysis provides a visible method which can be viewed, discussed and operated by individuals in the team. It is possible to reconsider ideas because the analysis follows a well-defined procedure which is documented and accessible. Although the process is systematic and disciplined, it still relies on the creative and conceptual ability of the analyst to determine meaning, salience and connections.

#### 11. Research Governance, Monitoring, Ethics and R&D Approval

Participants are not thought to be at risk by being involved in this study. The study will be conducted in accordance with approvals from NHS Tayside R&D and REC.

#### 12. Finance

The University of Dundee will fund the research as part of the Medical MBChB degree course.

#### 13. Indemnity

Under the requirements of the Scottish Executive Health Department's Research Governance Framework for Health and Community Care, the University of Dundee has agreed in principle to act as sponsor for this project. This provisional Sponsorship is subject to obtaining favourable ethical opinion and the appropriate local NHS R&D management approvals. The application number is 00000061.

#### 14. Report and Dissemination

The results will be presented as part of the Medical MBChB degree in the form of a fourth year project for Lisa Bryson. A summary of the study findings will be sent to all individuals expressing a wish to receive the results.

## References

1. Ciske, D.J., et al., Genetic Counseling and Neonatal Screening for Cystic Fibrosis: An Assessment of the Communication Process. *Pediatrics*, 2001. 107(4): p. 699-705.
2. Meldrum, C., C. Scott, and K.J. Swoboda, Spinal Muscular Atrophy Genetic Counseling Access and Genetic Knowledge: Parents' Perspectives. *Journal of Child Neurology*, 2007. 22(8): p. 1019-1026.
3. Rosas-Blum, E., P. Shirsat, and M. Leiner, Communicating genetic information: a difficult challenge for future pediatricians. *BMC Medical Education*, 2007. 7(1): p. 17.
4. Waitzkin, H., Doctor-Patient Communication: Clinical Implications of Social Scientific Research. *JAMA*, 1984. 252(17): p. 2441-2446.



Division of Medical Science

Centre for Oncology & Molecular Medicine

### Parents' experiences of genetic diagnosis in their child: an exploratory study

#### Participant Information Sheet

My name is Lisa Bryson and I am a fourth year medical student at Dundee University. I am required to undertake a project as part of my course and I invite you take part in the following study. However, before you decide to do so, I need to be sure that you understand firstly why I am doing it and secondly what it would involve if you agreed. I am therefore providing you with the following information. Please read it carefully and be sure to ask any questions you might have and, if you want, discuss it with others including your family and friends. I will do my best to explain the project to you and provide you with any further information you may ask for now or later.

#### Background Information

Very little is known about how parents are affected by having a child with a genetic condition. We want to find out how parents are affected by their child's diagnosis, what they understand about the diagnosis, what support the families may need and whether the age at which the child is diagnosed affects the parent's reaction. All parents will react differently and will need different amounts of support. We hope to find key points which need to be discussed when giving parents genetic counselling, advice and support for the future. The healthcare team want to be able to help parents as best they can.

#### Why have I been invited?

You have been invited to take part because you have a child who has a genetic condition. We hope to interview 6 people who have a child with a genetic condition.

#### What is involved?

If you decide that you want to take part in this study, it will involve meeting for one interview. You will be contacted by a researcher (Lisa Bryson) to arrange a convenient time and place for the interview. Interviews will take place in the clinical research centre in Ninewells, if convenient for you. If this was not possible, interviews may be held at the participants home. You will only be interviewed once, on a one to one basis, with Lisa Bryson and it will last a maximum of one hour. The questions will be about your memories of what happened and how you felt at the time of your child's diagnosis. The interview will be recorded; however the recordings will only be available to the researcher and supervisor. The recorded material will be analysed for the study to find out what experiences parents have at the time their child is diagnosed.

Taking part in this study is voluntary and entirely your decision. We will explain what the study involves and will answer any of your questions. You will need to sign a consent form indicating that you have agreed to take part; however you are free to withdraw from the study at any point. This will not affect any future involvement you may have with the clinical genetics team.

#### Expenses

As this is a student's research study, it will not be possible to reimburse any expenses.

#### What are the benefits of taking part?

The study is unlikely to benefit you directly. However, it may benefit parents of children with genetic conditions in the future. It is hoped that we will gain a better understanding of what parents need when their child is diagnosed with a genetic condition.

#### What are the risks in taking part?

Risks for taking part in this study are minimal. However, discussing personal experiences of a child's diagnosis may cause anxiety or distress. Further contact with the Clinical Genetics team will be available. Also, you might find attending an interview inconvenient. Steps will be taken to ensure that interviews are, as far as possible, held in a venue, and at a time, which is convenient for you.

#### Confidentiality: what will happen to the data collected in the project?

All information will be handled in confidence and the data we collect will not contain any personal information. This means that data in the study cannot be linked to your identity. All data will be stored on a password protected computer and only the researcher and supervisor will have access to the transcripts of the recordings for analysis. The results may be published, however this will not contain any information which would allow identification. Data will be kept securely for a maximum of 5 years, although all recordings will be destroyed following completion of the study.

At the end of the study you can ask to receive details of the findings. A summary of the results can be sent to you on completion of the project if you so wish.

#### What are my rights?

Participation in this study is entirely voluntary and you are free to refuse to take part or to withdraw from the study at any time without having to give a reason. Questions which participants do not feel completely comfortable in answering can be skipped and interviews can be stopped at any time if participants feel unable to continue.

If you believe that you have been harmed in any way by taking part in this study, you have the right to pursue a complaint and seek any resulting compensation through the University of Dundee who are acting as the research sponsor. Details about this are available from the research team.

#### Who Reviewed this study?

The Tayside Committee on Medical Research Ethics, which has responsibility for scrutinising all proposals for medical research on humans in Tayside, has examined the proposal and has raised no

objections from the point of view of medical ethics. It is a requirement that your records in this research, together with any relevant medical records, be made available for scrutiny by monitors from NHS Tayside, whose role is to check that research is properly conducted and the interests of those taking part are adequately protected.

What happens if there is a problem?

If you have concerns about the care you have received, you may wish to contact the Complaints and Advice Team, Level 7, Ninewells Hospital, Dundee, DD1 9SY

Freephone: 0800 027 5507; e-mail: [complaints.tayside@nhs.net](mailto:complaints.tayside@nhs.net)

Contact details for further information:

If you wish further information about the research or participation in the project, please contact Lisa Bryson who will be happy to answer any queries.

You can contact her by email at [L.J.Bryson@dundee.ac.uk](mailto:L.J.Bryson@dundee.ac.uk), by phone on 01382 496369 or send a letter to:

Lisa Bryson

c/o Jacqueline Dunlop  
Clinical Genetics,  
Level 6,  
Ninewells,  
Dundee  
DD1 9SY

Alternatively you can contact Dr Jonathan Berg, the project supervisor:

Jonathan Berg  
Clinical Genetics,  
Level 6,  
Ninewells,  
Dundee  
DD1 9SY  
[J.N.Berg@dundee.ac.uk](mailto:J.N.Berg@dundee.ac.uk)  
01382425716

Thank you for taking time to read this information sheet and considering participation in this project. If you are willing to take part in the project, please fill in the reply slip enclosed and return it in the free post envelope provided.

■

To be printed on NHS headed paper

Clinical Genetics,  
Level 6,  
Ninewells,  
Dundee,  
DD1 9SY

Dear <insert patient name here>

I am writing to ask if you would like to participate in a research study. We are hoping to find out more about how having a child diagnosed with a genetic condition has affected you. We would also like to find out what could help parents cope with this sort of information.

We are hoping to meet with you to ask you some questions about how your child was diagnosed, and how you felt about this. A patient information sheet, explaining more about the research study, can be found enclosed.

After reading about the study, if you are happy to take part, please return the reply slip enclosed, and we will contact you to make arrangements for the interview. If you prefer, you could contact Ms Jackie Dunlop on telephone number 01382496369

Alternatively, if you would simply like more information on the research study, please do not hesitate to get in touch, and we will be very happy to answer any of your questions.

Yours Faithfully,

Jonathan Berg

Consultant Clinical Geneticist



Division of Medical Sciences  
Centre of Oncology & Molecular Medicine

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Parents' experiences of genetic diagnosis in their child: an exploratory study

Interview Schedule

1. How old was your child at diagnosis? How and when was your child diagnosed?  
What was your initial reaction?
2. How much do you understand about of the condition? And the prognosis?
3. How much do you understand about future family planning for yourself? And  
your child?
4. How much did you understand about how the diagnosis would affect you  
financially, emotionally, socially and occupationally?
5. Do you think there are benefits of having a diagnosis?
6. How has the family been affected by the diagnosis? Including siblings?
7. What/who helped in coming to terms with the diagnosis and what was  
unhelpful?
8. What advice would you give to parents of a newly diagnosed child?



### East of Scotland Research Ethics Service

#### Tayside Committee on Medical Research Ethics A

Research Ethics Office  
TAHSC, Residency Block C  
Ninewells Hospital & Medical School  
DUNDEE  
DD1 9SY

Miss Lisa J Bryson  
c/o Clinical Genetics  
Level 6, Ninewells Hospital  
DUNDEE  
DD1 9SY

Date: 13 January 2011  
Your Ref:  
Our Ref: LR/10/S1401/64  
Enquiries to: Mrs Lorraine Reilly  
Extension: Ninewells extension 40099  
Direct Line: 01382 740099  
Email: lorraine.reilly@nhs.net

Dear Miss Bryson

**Study Title:** Parents' experiences of genetic diagnosis in their child: an exploratory study  
**REC reference number:** 10/S1401/64  
**Protocol number:** 00000061

Thank you for your letter of 10 January 2011. I can confirm the REC has received the documents listed below as evidence of compliance with the approval conditions detailed in our letter dated 17 December 2010. Please note these documents are for information only and have not been reviewed by the committee.

#### Documents received

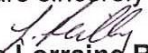
The documents received were as follows:

Document	Version	Date
Covering Letter		10 January 2011
Participant Information Sheet	1.2	05 January 2011
Participant Consent Form	1.2	05 January 2011

You should ensure that the sponsor has a copy of the final documentation for the study. It is the sponsor's responsibility to ensure that the documentation is made available to R&D offices at all participating sites.

<b>10/S1401/64</b>	<b>Please quote this number on all correspondence</b>
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Yours sincerely

  
**Mrs Lorraine Reilly**  
Coordinator



## College of Medicine, Dentistry and Nursing Division of Clinical & Population Sciences & Education

*Director of Clinical & Population Sciences & Education*

Professor Frank Sullivan PhD FRCP FRCGP

10 October 2010

APP No 00000061

Lisa Bryson  
Medical Student  
Ninewells Hospital & Medical School  
Dundee  
DD1 9SY

Project Title: How the timing of the diagnosis of a genetic condition in a child affects their parents.

Dear Ms Bryson

### **Sponsorship of Project**

Under the requirements of the Scottish Executive Health Department's Research Governance Framework for Health and Community Care, the University of Dundee agrees in principle to act as sponsor for this project. This provisional Sponsorship is subject to you obtaining a favourable ethical opinion and appropriate local University & NHS R&D management approvals (if required).

As Chief Investigator, you must remember that you must not begin your project until all necessary approvals have been obtained. If the details of the project change, either during the process of obtaining these approvals, or during the project itself, you must notify Dr Davis immediately, and if appropriate the Ethics committee, and the relevant R&D offices.

Lastly, please make sure that you quote the Application Number given above in any correspondence.

Yours sincerely

Dr Julian P L Davis □ Lecturer, Division of Clinical & Population Sciences & Education  
t 01382 420104

e [j.p.l.davis@cpse.dundee.ac.uk](mailto:j.p.l.davis@cpse.dundee.ac.uk)

UNIVERSITY OF DUNDEE · The Mackenzie Building, Kirsty Semple Way, Dundee DD2 4BF  
Scotland UK t +44 (0) 1382 420000 e [info@cpse.dundee.ac.uk](mailto:info@cpse.dundee.ac.uk) □ Scottish Charity No. SC015096

<b>Welcome to the Integrated Research Application System</b>
<b>IRAS Project Filter</b>
The integrated dataset required for your project will be created from the answers you give to the following questions. The system will generate only those questions and sections which (a) apply to your study type and (b) are required by the bodies reviewing your study. Please ensure you answer all the questions before proceeding with your applications.
<b>Please enter a short title for this project</b> (maximum 70 characters) Parents' experiences of genetic diagnosis in their child
<b>1. Is your project research?</b>  <input checked="" type="radio"/> Yes <input type="radio"/> No
<b>2. Select one category from the list below:</b>  <input type="radio"/> Clinical trial of an investigational medicinal product <input type="radio"/> Clinical investigation or other study of a medical device <input type="radio"/> Combined trial of an investigational medicinal product and an investigational medical device <input type="radio"/> Other clinical trial or clinical investigation <input type="radio"/> Study administering questionnaires/interviews for quantitative analysis, or using mixed quantitative/qualitative methodology <input checked="" type="radio"/> Study involving qualitative methods only <input type="radio"/> Study limited to working with human tissue samples, other human biological samples and/or data ( <i>specific project only</i> ) <input type="radio"/> Research tissue bank <input type="radio"/> Research database  <b>If your work does not fit any of these categories, select the option below:</b> <input type="radio"/> Other
<b>2a. Please answer the following question(s):</b>  a) Does the study involve the use of any ionising radiation? <input type="radio"/> Yes <input checked="" type="radio"/> No b) Will you be taking new human tissue samples (or other human biological samples)? <input type="radio"/> Yes <input checked="" type="radio"/> No c) Will you be using existing human tissue samples (or other human biological samples)? <input type="radio"/> Yes <input checked="" type="radio"/> No
<b>3. In which countries of the UK will the research sites be located?</b> ( <i>Tick all that apply</i> )  <input type="checkbox"/> England <input checked="" type="checkbox"/> Scotland <input type="checkbox"/> Wales <input type="checkbox"/> Northern Ireland  <b>3a. In which country of the UK will the lead NHS R&amp;D office be located:</b>  <input type="radio"/> England <input checked="" type="radio"/> Scotland

<input type="radio"/> Wales <input type="radio"/> Northern Ireland <input type="radio"/> This study does not involve the NHS
<b>4. Which review bodies are you applying to?</b> NHS/HSC Research and Development offices Research Ethics Committee National Information Governance Board for Health and Social Care (NIGB) Ministry of Justice (MoJ) National Offender Management Service (NOMS) (Prisons & Probation)
<b>5. Will any research sites in this study be NHS organisations?</b> <input checked="" type="radio"/> Yes <input type="radio"/> No
<b>6. Do you plan to include any participants who are children?</b> <input type="radio"/> Yes <input checked="" type="radio"/> No
<b>7. Do you plan at any stage of the project to undertake intrusive research involving adults lacking capacity to consent for themselves?</b> <input type="radio"/> Yes <input checked="" type="radio"/> No <i>Answer Yes if you plan to recruit participants aged 16 or over who lack capacity, or to retain them in the study following loss of capacity. Intrusive research means any research requiring consent in law. This includes use of identifiable tissue samples or personal information, except where application is being made to the NIGB Ethics and Confidentiality Committee to set aside the common law duty of confidentiality in England and Wales. Please consult the guidance notes for further information on the legal frameworks for research involving adults lacking capacity in the UK.</i>
<b>8. Do you plan to include any participants who are prisoners or young offenders in the custody of HM Prison Service or who are offenders supervised by the probation service in England or Wales?</b> <input type="radio"/> Yes <input checked="" type="radio"/> No
<b>9. Is the study, or any part of the study, being undertaken as an educational project?</b> <input checked="" type="radio"/> Yes <input type="radio"/> No
<b>9a. Is the project being undertaken in part fulfilment of a PhD or other doctorate?</b> <input type="radio"/> Yes <input checked="" type="radio"/> No
<b>10. Will this research be financially supported by the United States Department of Health and Human Services or any of its divisions, agencies or programs?</b> <input type="radio"/> Yes <input checked="" type="radio"/> No

NOTICE OF SUBSTANTIAL AMENDMENT	
<p>Please use this form to notify the main REC of substantial amendments to all research other than clinical trials of investigational medicinal products (CTIMPs). For CTIMPs, please use the European Commission notice of substantial amendment form at <a href="http://eudract.emea.europa.eu/document.html">http://eudract.emea.europa.eu/document.html</a>.</p> <p>The form should be completed by the Chief Investigator using language comprehensible to a lay person. Support in principle should be sought from the study sponsor before the amendment is submitted.</p>	
<p><b>Details of Chief Investigator:</b></p>	
	<p>Title Forename/Initials Surname</p> <p>Dr Jonathan Berg</p> <p>Work Address Clinical Genetics</p> <p>Level 6, Ninewells Hospital</p> <p>Dundee</p>
<p><b>Full title of study:</b> Parents' experiences of genetic diagnosis in their child: an exploratory study</p>	
<p><b>Lead sponsor:</b> University of Dundee</p>	
<p><b>Name of REC:</b> Tayside A</p>	
<p><b>REC reference number:</b> 10/1401/64</p>	

or a document listing the changes and giving both the previous and revised text.

Protocol Submitted

(c) Amendment to the information sheet(s) and consent form(s) for participants, or to any other supporting documentation for the study

☒ Yes ☐ No

If yes, please submit all revised documents with new version numbers and dates, highlighting new text in bold.

Questionnaire has been submitted

**Is this a modified version of an amendment previously notified and not approved?**

☐ Yes ☒ No

### Summary of changes

Briefly summarise the main changes proposed in this amendment. Explain the purpose of the changes and their significance for the study.

If this is a modified amendment, please explain how the modifications address the concerns raised previously by the ethics committee.

If the amendment significantly alters the research design or methodology, or could otherwise affect the scientific value of the study, supporting scientific information should be given (or enclosed separately). Indicate whether or not additional scientific critique has been obtained.

As mentioned in the previous amendment, a questionnaire has now been developed from the information gained in the interviews. It has been developed for the consultant paediatricians and geneticists within Tayside. These consultants will be involved in the management of families affected by a genetic diagnosis. The questionnaire aims to find out how often key themes found from the interviews are discussed within consultations. This will help highlight whether a management framework should be developed to ensure parents are receiving all of the information and follow up which they require at the time of diagnosis.

11 interviews were done with parents of children with genetic diagnoses. These 11 interviews showed that there is a need for further thought to be put into how these families are managed by the healthcare team. Parents discussed difficulties accessing the support which is available and a lack of sign posting and sufficient appropriate information being provided.

These results will be important with the advent of micro-array testing, as more and more parents are given genetic diagnoses for their children. There is a need to document this information as there are a limited number of research documents in the literature on this subject.

Currently every regional research centre is taking part in the Deciphering Developmental Delay (DDD) study which is looking at genetic testing in learning disability. I may have the opportunity to disseminate my study results to this research forum. My research will hopefully help understand how to manage the parents and families of children who are given genetic diagnoses.

### Any other relevant information

Applicants may indicate any specific issues relating to the amendment, on which the opinion of a reviewing body is sought.

### List of enclosed documents

Document	Version	Date
Protocol	1.4	30/05/2012
Questionnaire	1.1	30/05/2012

### Declaration by Chief Investigator

1. *I confirm that the information in this form is accurate to the best of my knowledge and I take full responsibility for it.*
2. *I confirm that the study sponsor has been notified of the proposed amendment.*
3. *I consider that it would be reasonable for the proposed amendment to be implemented.*

This section was signed electronically by Jonathan Berg on 06/06/2012 13:40.

Job Title/Post: Consultant clinical geneticist

Organisation: Nhs tayside

Email: J.n.berg@dundee.ac.uk

Date:.....

## Parents' experiences of genetic diagnosis in their child: an exploratory study

I am Lisa Bryson, a final year medical student, and I am currently undertaking a part time MSc in research. My research topic is on parents' experiences of having a genetic diagnosis for their child who is developmentally delayed; either motor, social, language or global. I have interviewed 11 mothers and 1 father asking them about their experiences.

For the next part of my research, I want to ask paediatricians about the management of children with a genetic diagnosis causing developmental delay.

If you would be able to complete this questionnaire and return it to me in the envelope provided, it would be appreciated.

Please "X" the relevant box for each of these questions

1. What is your medical speciality?

General Paediatrician

☐

Neonatologist

☐

Geneticist

☐

Community Paediatrician

☐

2. How often do you see children who have a diagnosed genetic condition causing developmental delay?

Weekly

☐

Monthly

☐

Every few months

☐

Rarely

☐

For the following questions, please answer them in relation to managing a child who is diagnosed with a genetic condition causing developmental delay

	Always	Often	half of the time	Sometimes	Never
3. Do you screen for depression in the parent(s)?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
4. Do you ask the parent(s) about the coping strategies of other family members, such as the grandparents and/or siblings of the child with the diagnosis?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
5. Do you ask the parent(s) if they have read any research on the diagnosis?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
6. If research has been read, do you offer to discuss it with the parent(s)?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
7. Do you ask the parent(s) if they feel they are getting sufficient support from the healthcare team?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

- |     |  |                                    |                                   |   |                                       |                                   |
|-----|--|------------------------------------|-----------------------------------|---|---------------------------------------|-----------------------------------|
|     |  | Always                             | Often                             | half of the<br>time                             | Sometimes                             | Never                             |
| 8.  | Do you ask the parent(s) if they have accessed the financial support available to them?  | <input type="checkbox"/>           | <input type="checkbox"/>          | <input type="checkbox"/>                        | <input type="checkbox"/>              | <input type="checkbox"/>          |
| 9.  | Would you feel confident delivering advice on available financial support?   | Yes<br><input type="checkbox"/>    | No<br><input type="checkbox"/>    |   |                                       |                                   |
| 10. | Do you ask the parent(s) if they have accessed available associations which may or may not be specific to the condition which their child has? | Always<br><input type="checkbox"/> | Often<br><input type="checkbox"/> | half of the<br>time<br><input type="checkbox"/> | Sometimes<br><input type="checkbox"/> | Never<br><input type="checkbox"/> |
| 11. | Would you feel confident to give the parent(s) advice on associations available to them?   | Yes<br><input type="checkbox"/>    | No<br><input type="checkbox"/>    |   |                                       |                                   |
| 12. | Do you ask the parent(s) if they have childcare available for the child with the diagnosis?  | Always<br><input type="checkbox"/> | Often<br><input type="checkbox"/> | half of the<br>time<br><input type="checkbox"/> | Sometimes<br><input type="checkbox"/> | Never<br><input type="checkbox"/> |
| 13. | Would you feel confident to give the parent(s) advice on childcare available for the child with the diagnosis?                                 | Yes<br><input type="checkbox"/>    | No<br><input type="checkbox"/>    |   |                                       |                                   |
| 14. | Do you ask the parent(s) if they have information on schooling for their child with the diagnosis?   | Always<br><input type="checkbox"/> | Often<br><input type="checkbox"/> | half of the<br>time<br><input type="checkbox"/> | Sometimes<br><input type="checkbox"/> | Never<br><input type="checkbox"/> |
| 15. | Would you feel confident to give the parent(s) advice on schooling for their child with the diagnosis?   | Yes<br><input type="checkbox"/>    | No<br><input type="checkbox"/>    |   |                                       |                                   |
| 16. | Do you ask parent(s) if they have information on future reproductive choices?  | Always<br><input type="checkbox"/> | Often<br><input type="checkbox"/> | ½ of the<br>time<br><input type="checkbox"/>    | Sometimes<br><input type="checkbox"/> | Never<br><input type="checkbox"/> |
| 17. | Would you feel confident to give the parent(s) advice on future reproductive choices?  | Yes<br><input type="checkbox"/>    | No<br><input type="checkbox"/>    |   |                                       |                                   |



### Questionnaire development

After the eleven interviews, a questionnaire was developed for the consultant paediatricians and geneticists within Tayside based on the themes found in the interviews. The results of the questionnaire will help us to determine how often the key themes identified in the interviews are discussed within consultation. This will help highlight whether a management framework should be developed to ensure parents are receiving all of the information and follow up which they require at the time of diagnosis.

Validation of the questionnaire was done in a way that was in keeping with necessary development techniques. The questionnaire was developed for the completion by the specialists who are involved in the management of the children and parents of children who are affected by a genetic diagnosis. It was short in length so that it could be completed quickly and with ease. Before being tested for validity and reliability, the clarity and face validity of the questionnaire was checked by colleagues. This provided information on approximate timing to read through the questionnaire and ensured the language was easy to understand.

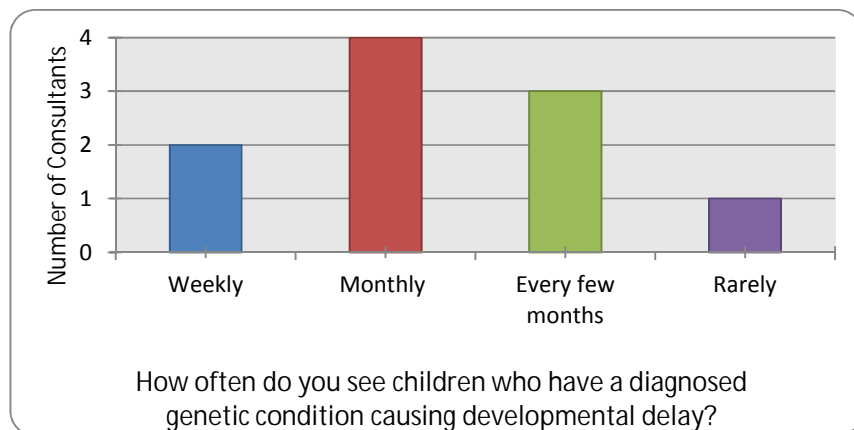
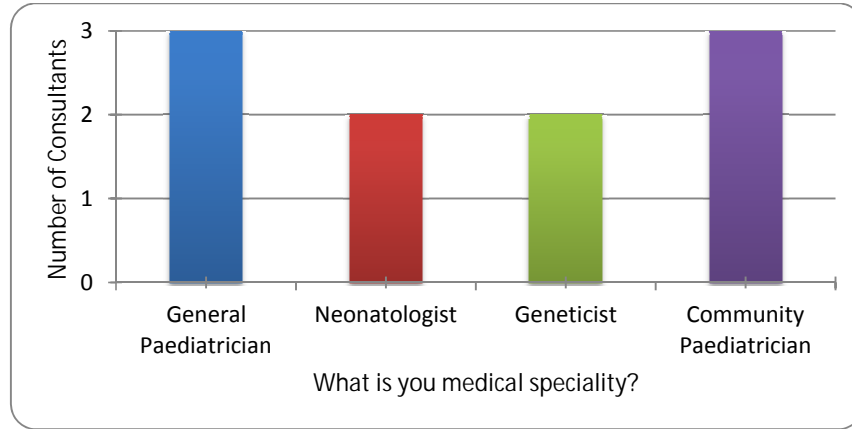
Content validity was ensured as specialists in the area of genetics and genetic counselling were asked to assess the questionnaire. The advice given to me was taken into account and the questionnaire was adapted.

Reliability testing was not checked due to time constraints and the small size of the study.

### Questionnaires Results

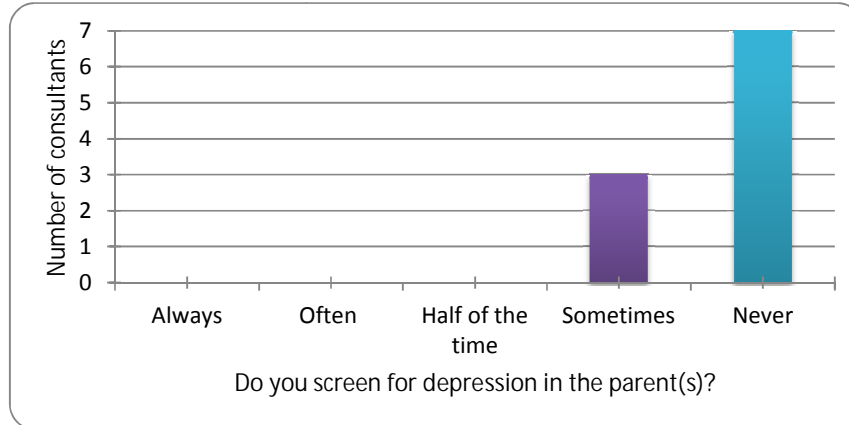
The questionnaires were used as a tool to find out how often consultant paediatricians involved in the management of these patients think they cover the main themes, which were identified within the interviews. A range of different consultants, who see children who have a diagnosed genetic condition, a variable amount, completed the questionnaire. The results suggest that there may be a need for a more systematic approach when developing a management plan for parents who have a child diagnosed with a genetic

condition, as none of the key themes identified were thought to be picked up “always” by the consultants.



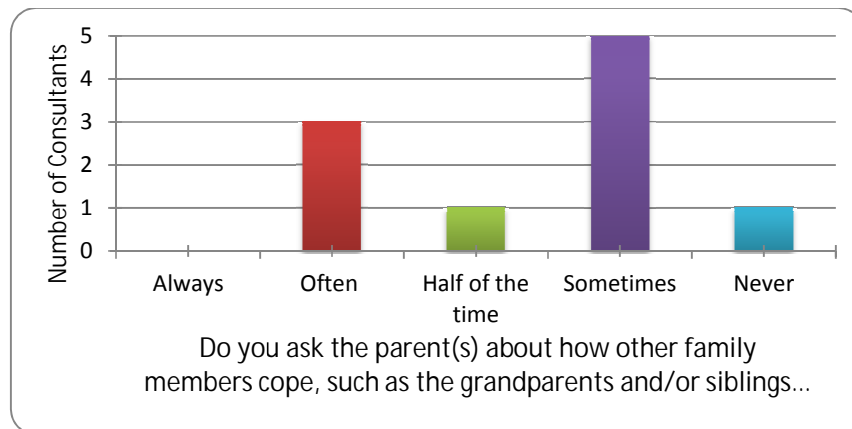
### Postnatal depression

Previous research has found that depression can be common in parents of children with a variety of genetic disorders (19, 26, 33). The interviews correlate with this as a number of parents described suffering from postnatal depression, which they associated with the condition that their child suffered from. In relation to this, when looking at the results of the questionnaires it can be seen that 7 of the 10 consultant paediatricians “never” screened for depression in the parents and 3 “sometimes” did. Mother 2 described being seen by many different healthcare professionals yet there being a delay in having her postnatal depression diagnosed. This delay could have been prevented if doctors managing her child had considered screening for depression.



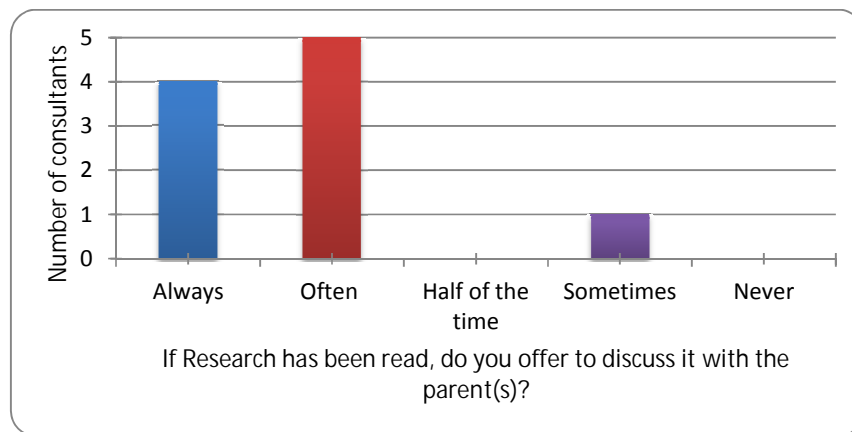
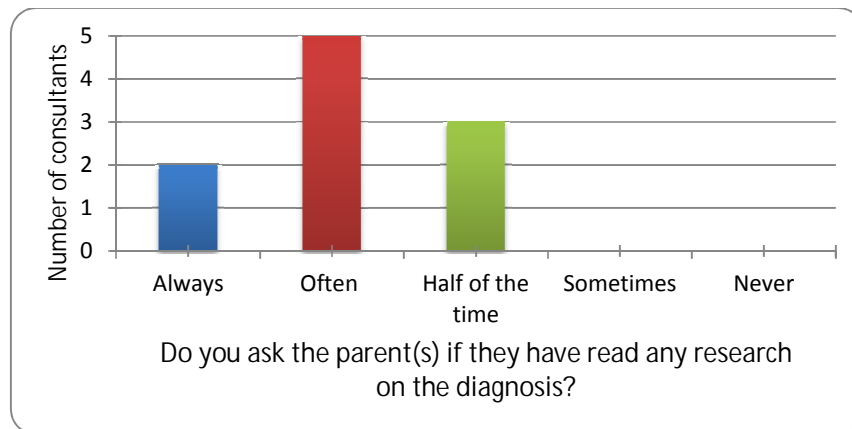
### Other family members

During the interviews, some of the parents discussed the difficulties involved in explaining the condition, which their child suffers from, to other family members, in particular grandparents and siblings of the child. These family members may also experience grief and require support and relevant information. Mother 5 had gained access to a course in which she was able to gain knowledge on how to discuss the diagnosis with her children. Mother 6 had doctors within her family who were able to provide help explaining information. Some of the other parents mentioned feeling they would have benefitted from similar resources, general advice or leaflets for family members. The results of the questionnaires correlate with these reports as none of the consultants felt they asked about how family members were coping “always”. This suggests that access to support for other family members is not consistent.



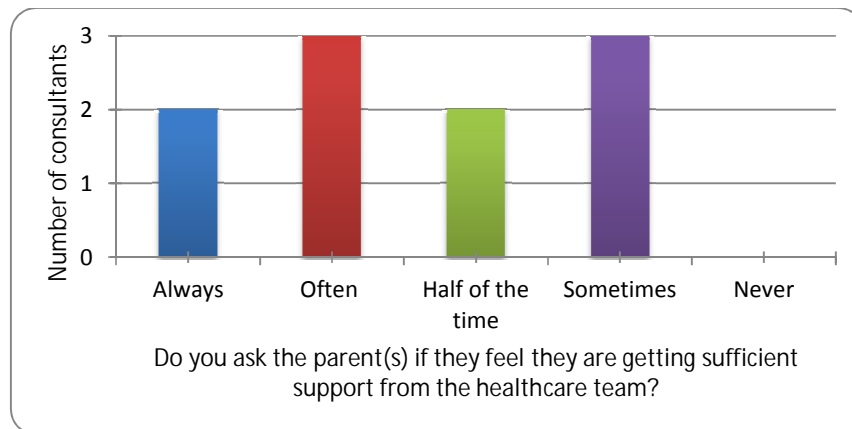
### Research

The information which parents are given can have an impact on the parents' initial experiences of genetic diagnosis. Parents described finding "worst case scenarios" whilst doing their own research on-line. All of the parents agreed that using the Internet and doing research without direction could have negative repercussions. In the questionnaires, consultant paediatricians were questioned on whether they ask parents about the research they have read and discuss it with them. All consultants felt they asked if parents read research at least "half of the time". Additionally, if research was read the majority of consultants said they "often" or "always" asked the parents if they want to discuss it. However, parents described this as an area in which they did not always feel supported. Parent 8, in particular, discussed reading research and not being given time to discuss what she had found. In order to ensure parents feel they have a good understanding of their child's condition and are not confused by information they have read, research should be discussed during consultations.



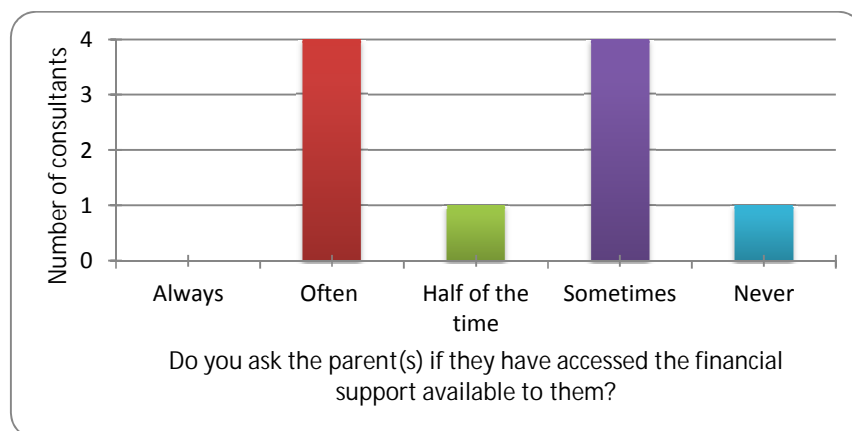
### Support from the healthcare team

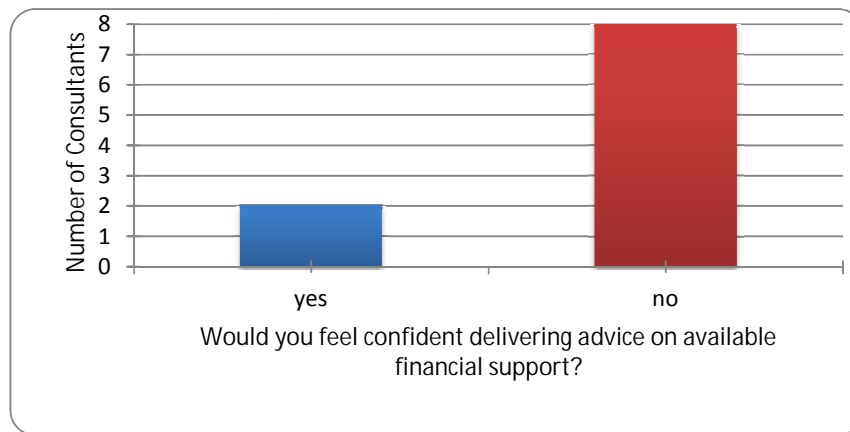
Access to support was one of the main areas of complaint from the parents, who wanted to know what support was available to them and how to gain access to it. Within the questionnaires, the consultants were asked how often they discuss parents' access to support. There was a spread in the results, from 'sometimes' to 'always'. None of the consultants answered 'never'. The solution for preventing parents from feeling unsupported may be to ensure access to support is always discussed. Alternatively, a more systematic approach to providing support, follow-up and information on what is available to parents could alleviate parents' criticisms.



### Financial support

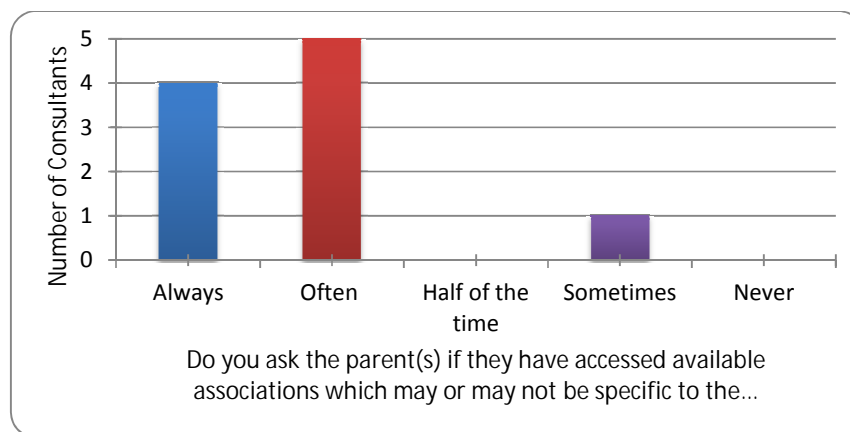
Financial support would have been available for all of the parents who took part in the interviews. Difficulties in accessing it were identified and described by the parents, for example not knowing their eligibility or how to access it and having difficulty completing the paperwork were mentioned. From the questionnaires, none of the consultants claimed to discuss financial support “always” and the vast majority did not feel confident to provide the advice. This identifies a need for education to be provided to doctors on financial support and where to direct parents to for help. This would hopefully prevent families missing out on finances available to them.

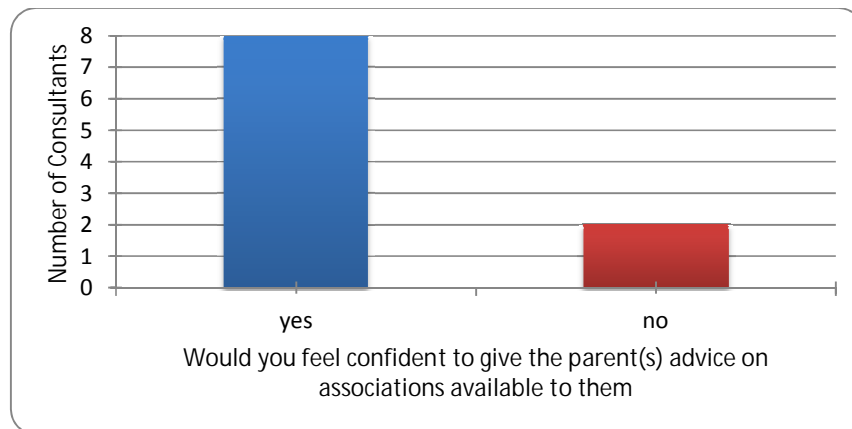




### Association

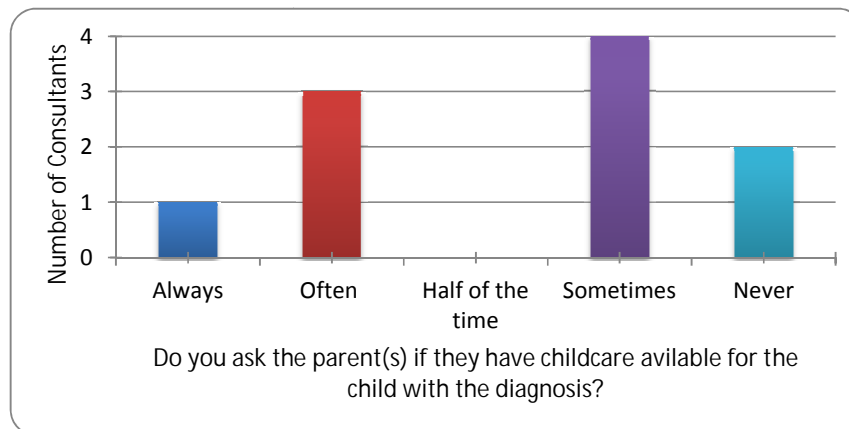
In contrast to financial support, the majority of consultants, who completed the questionnaire, felt confident to supply information on support groups and felt they gave advice the majority of the time. However, this did not always appear to be the case in interviews. Mother 1 would have liked to have been directed to a support group but did not know where to turn. There may be a problem in the communication between some consultants and parents, when it comes to providing information on available associations and support groups.



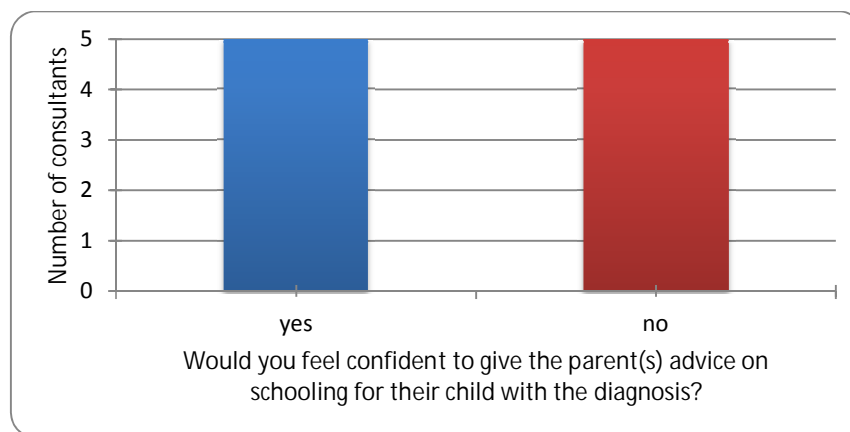
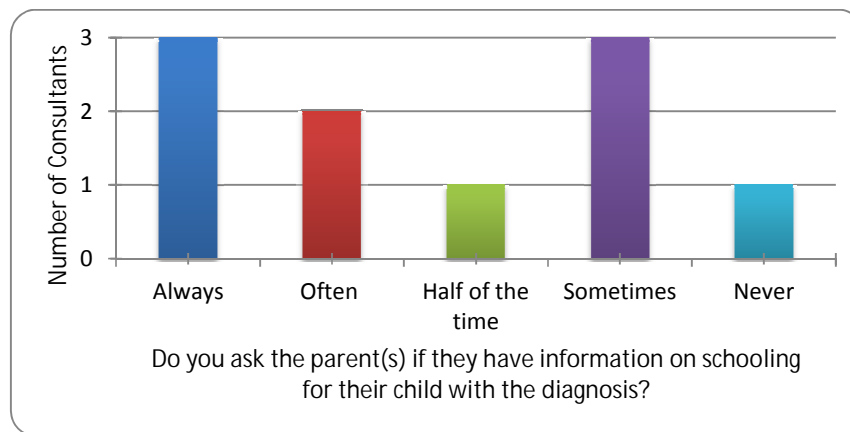
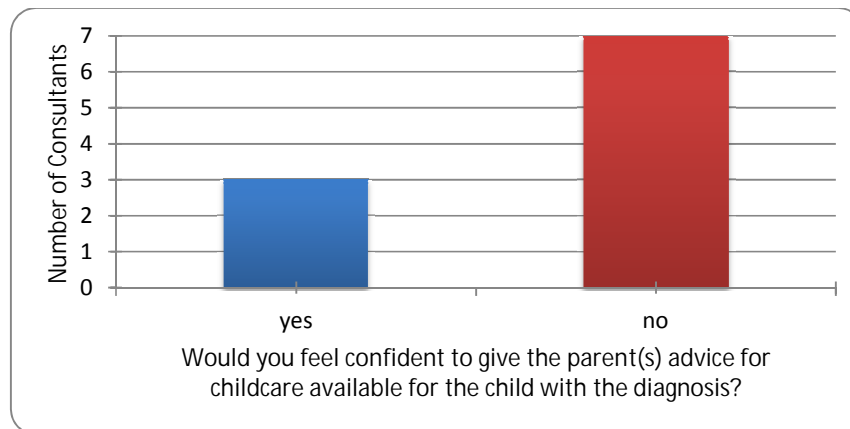


### Childcare and schooling

Childcare and schooling for children with additional needs appeared to cause the parents stress. This was in relation to ensuring their child's needs were communicated and managed appropriately, knowing what childcare and schools were available and suitable, and knowing how to manage transitions between childcare, nurseries and schools. A few of the parents were offered advice on schooling and childcare facilities, whereas others were not. This may relate to the split that can be seen in the consultants' answers when they were asked whether they discuss childcare and schooling with parents. The majority of consultants admitted they do not feel confident to provide advice on childcare and there was a 50/50 split as to whether the consultants felt confident to provide advice on schooling. Some of the consultant neonatologists documented that discussions about schools were not relevant to them as they deal with children at a much younger age.



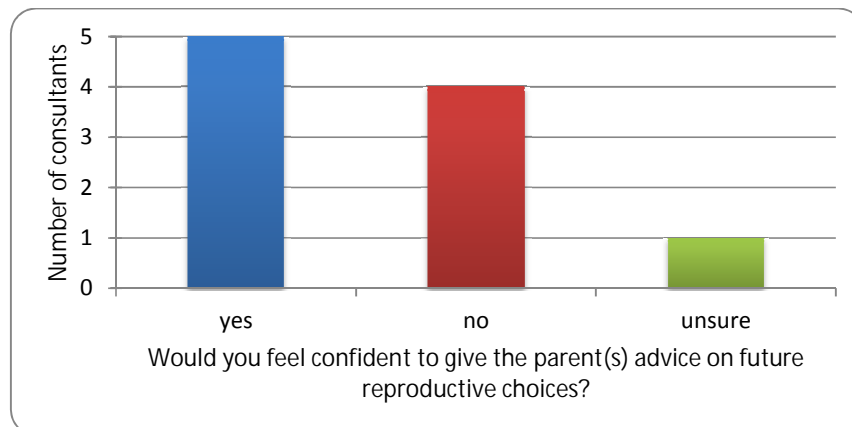
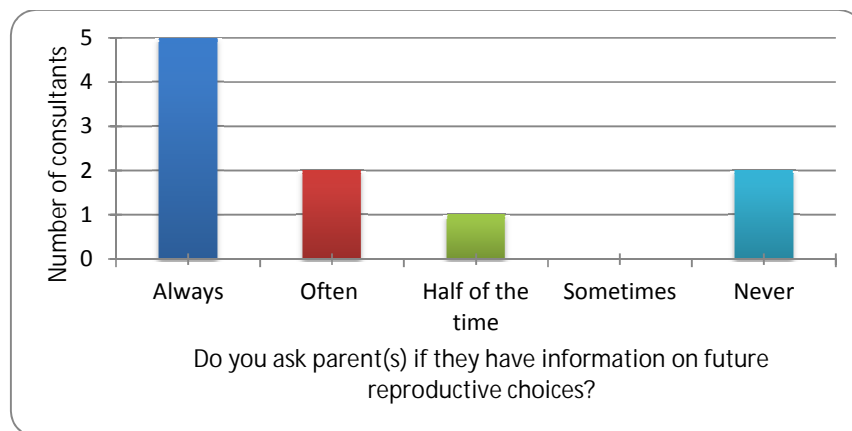




### Reproductive choices

The parents generally felt they gained a good understanding of how the genetic condition had developed and what their risks were for future pregnancies. Therefore this is something

that the healthcare professionals must explain relatively well. In the questionnaires, there was a split between the specialities. Neonatologists and geneticists all said “always” to the question “Do you ask parent(s) if they have information on future reproductive choices?”. Although this is a very small study this may suggest that these specialities are very good at providing this type of information. If the other themes were also discussed “always” by a certain speciality then this would ensure that certain topics were not missed from some parents. In this study, mother 4 was able to use the diagnosis to make a decision on family planning and use IVF to ensure her next child would not be affected by the same condition. Highlighting how important consideration of future family planning was for her family.



### Limitations of the Questionnaires

There were only 10 questionnaires completed. There is a multi disciplinary team involved in the care of a child with additional needs and a genetic diagnosis. Other healthcare professionals and non-healthcare professional could have been involved in the questionnaire to determine who provides what information.

## Additional Quotes for each theme

The reaction to having a genetic diagnosis included:

- Parents' experiences of not knowing the diagnosis

"time went on and on and on and most of the results had come back negative we'd got that back, we'd been told that everything had been coming back negative and we thought oh good we can breathe it's CP. So it was a bit of a shock when we got the HSP diagnosis because we had kind of talked ourselves out of it being HSP"- Interview 5

"they mentioned that they thought it might be Edwards syndrome which was really difficult to kind of take on.... So when they said they've not found anything it's \*\*, I'd already looked into all this on the internet before hand and this was one diagnosis that I thought well it's more hopeful than anything else that has been said, so when they said that I was like uh, if that's all you can say and you can't find anything else then that's I was relieved."- Interview 10

"It was going through that grieving process again. But it was easier because I knew what was happening. I knew what was happening in my head and I knew I was going through that overwhelmed feeling and the bit of anger feeling and then the I can't do anything about this feeling. You know it's just that grieving process that you go through and that was quite hard. And actually quite short although saying that it's still here now. It's still under the covers"- interview 5

"having the change of diagnosis ...was a bit scary because [condition] seemed to be so much more profoundly disabled and didn't live as long as the original bracket that he was under."- Interview 6

"I suppose, I don't really sit and kind of ask myself why...I've never really done that because I'm not going to, I'm not going to find an answer to it ... I guess if they could, maybe it would sort of give it a bit more closure to it. I don't know if it would or not ... if it's not going to add anything to I guess her management of her difficulties then you know I don't know."- Interview 10

"I knew from the day he was born that there was something wrong with him. Just the mother's instinct. I cried and cried and cried as I was obviously distraught. And I couldn't put my finger on it"- Interview 4

"I was adamant probably from the day he was born that there was something not quite right and everybody else kept on going "no, no he's fine, he's just a little bit slow, he's ok" so I'd spent so long trying to convince myself that there was nothing wrong with him and I think I'd just convinced myself that there was nothing wrong and then he was diagnosed"- interview 9 mum

"there is nothing worse than being in limbo I just couldn't do that. WE know YY goes to nursery with another little girl whose 3 months older than him and she has severer learning disabilities and they don't know what it is. And I think that must be a nightmare. Again I think it's the not knowing what is going to come, not knowing if they are going to grow out of it or if they are going to be able to crawl or roll over or walk or speak, that would be a lot worse than knowing what's coming, and being able to plan your life out and get on with it."- Interview 9 mum

"we worried because we didn't really know what was wrong with her. You do wonder if it's some life limiting disease. But luckily this one isn't. But up until she was about 2 and a half we really didn't know what to expect. We didn't know if we would have her for much longer. It ... So it was actually quite good to get the proper diagnosis, we were actually quite ecstatic... (Pre- diagnosis) I was a bit standoffish, because I just didn't know, that sounds awful as a mother, we did bond, but I kind of felt I don't know how long I'm going to have her. I don't, you just didn't want to invest your heart and soul in her because you just didn't know if she was going to live."- interview3

"From the start I very much thought it was from me because of my epilepsy....had made me feel a little bit guilty...now I don't think it has come from either of us. That makes me feel a bit better, that it's not been my fault."- Interview 7

"it would be surprising if it wasn't ...I'm pretty positive that it is. ...But I've been in touch with professor ... as well, in Germany, and e-mailed over the clinical photographs of YY and he said definitely ... Without question, without even seeing him or looking at his medical notes or anything...it will be good to know for definite. I never thought they'd ever identify the gene that caused it. I just thought that it would be, "oh well he's probably got that" and that is fine. But for them to actually have identified the gene is quite good and to have a definite, 100% diagnosis."- Interview 6

"So four months basic limbo, not knowing what the matter was"- Interview 8

- Receiving the diagnosis

"me and my husband stood on the concourse crying our eyes out because we didn't know where to, we just felt at that time there was nobody to kind of"- interview 8

"HSP breaking bad news, that was a lot better because I knew (paediatrician), (paediatrician) had been with us as a family for 4 years and she knew my husband she knew that we had AA and she knew YY and so that really helps and I could just be myself with her, I could just, I think one of the things I said was oh god AA, and then I went blah blah blah all over the place and it was good because I just happened to be in Dundee which meant she could do it face to face. It was so much better than getting the phone calls."- interview 5

"you were upset so I was trying to be the male and be the stronger one and say it's alright. Kind of that attitude towards it obviously still thinking about it in my head... (she) more upset than I was and my attitude was right just get on with it."- dad interview 9

"I can be adamant that something bad is going to happen and you'll be adamant that something good is going to happen and I, you don't let me persuade you that it might not go good and I don't let you persuade me that it won't."- Interview 9 mum

"I don't know what's happening about that to run micro RA something, some complicated thing with all the genes"- interview 10

"I just remember there was times we would sort of come in and there would be people around his cot and it was almost like –who are you?" - Interview 8

"basically this is the syndrome that he's got, wrote it down on a piece of paper and said I don't actually know much about this syndrome, go look it up on the internet and you'll find out" - Interview 8

"her general sort of outlook was going to be a struggle to start, she was going to struggle to thrive for the first sort of 6 months to a year. And then things should get a lot easier...It made me think you know after 6 months everything will be fine." - interview 1

"I thought we were being told she might die basically. That's all I can remember" - interview 1

- The benefits and drawbacks of having a diagnosis

"It was just a name floating" - interview 9 mum

"it is a rubber stamp that your child is always going to have special needs... you always kind of had that hope that it might just be a temporary blip and when he's 1 he'll catch up. But he's not." - Interview 8

"I'm sure there are lots of hysterical mothers but you know "ah, there is something wrong", why would you take them seriously without a bit more to it than that...when I was being told that it was my fault, the implication that I was a sort of bad mother, you're not feeding your child properly. It all comes down to not being able to provide his most basic needs and he is going to have to go into SCBU because he is failing to thrive because you're not feeding him properly... And if I wasn't then that was, the proof is in the pudding, my oldest is there. I thought that her developmental pattern is normal and I breast fed her for 13-14 months. ... she did everything and she was a high flyer. And I just thought that was normal, every child did that. So you know, the implication to me was that I wasn't doing what I was meant to be doing as a mother, as a parent. So, to get justification that it wasn't my failings just made me feel a bit better in a way. I mean obviously you'd rather not have the diagnosis" - Interview 4

"you'd be just kind of stabbing about in the dark, not knowing where you were going without kind of, well I suppose you'd just kind of take it as it comes but it's helpful to be able to forward plan. Just seeing how other children have coped although I know they're all different and there is quite a wide spectrum. It does give you a rough idea as to what to expect." - Interview 3

"It was good to like say to like my mum or my grandparents or whatever, oh he's got this particular syndrome rather than we just don't know what's wrong with him. So it was useful in that respect" - interview 6

"every one very excited around us and they told us that he had \*\* syndrome which seemed a bit bizarre ... I just kind of remember that day, "we've got some good news for you" and sort of a lot of that and then we got taken into our appointment and that's when they told us" - Interview 8

"but when we got the diagnosis I obviously went straight and looked everything up on the internet and it was all quite pessimistic and not very good news. So you can think getting a diagnosis isn't necessarily a good thing from my point of view." - Interview 6

"it was a bit that from a clinical point of view, I can see this is very positive outcome but from a parental point of view it's not, it's quite hard and that must be a very difficult balance to have because you can see they go woo we've got a positive, when actually we don't want to hear that positive part of it. It's not a positive for us and that kind of must be quite difficult because medicine wants to put labels on things and sometimes as a parent you don't want to have those labels put on it" - Interview 5

"it makes it easier to accept it because it's there, whereas I suppose more with having an autistic child to a certain degree, it's the amount of opinion as well." - interview 8

"I'm concerned with a label" - parent 1

"if I was just told there was a problem then you're going to end up worrying even more because you want to know" - parent 2

"Even though it meant, it was going to bring a lot more problems with it but, rather knowing than not knowing yeah." - parent 2

"right away, we went home that day having spoken to the geneticist and we looked it up online and there was a picture of a child and that was her." - parent 3

"Whereas if it wasn't diagnosed as Williams syndrome you would wonder why isn't she walking now why cant she crawl and that kind of thing." - Interview 11

"you don't just want to be wandering; you know what is it what's wrong." - Interview 7

"It's really amazing to see photographs of these other kids and they all look very similar but that's about it really" - Interview 6

"Yeah it will be good to know for definite. I never thought they'd ever identify the gene that caused it. I just thought that it would be, "oh well he's probably got that" and that is fine" - interview 6

"up until she was about 2 and a half we really didn't know what to expect. We didn't know if we would have her for much longer. It sounds quite dramatic. So it was actually quite good to get the proper diagnosis, we were actually quite ecstatic" – interview 3

- The emotional reaction to the diagnosis

"I was putting a front on it and I didn't want to admit to people that inside I was slightly heartbroken but you just want to kind of seem like a strong person ...Like I've said before, I don't know if when YY goes to school and I've got time on my hands, maybe everything will come crashing down and maybe

I'm not dealing with things a lot more than I like to portray or things like that but at the moment everything is fine...everything might come crashing down but today he's my little boy and that's what life is for him so you just get on with it"- interview 8

"you almost grieve for the child that you thought that you gave birth to but then you kind of regroup and reassess and realise they are still you're children, it's nothing they've done. They're still wonderful gorgeous people"- parent 4

"it did take a while to get around your child is not going to grow up as you would love your children to grow up"- Interview 5

"That's the bit that is kind of hard when you see it. And we got a letter from YYs orthopaedic consultant that he saw in clinic a couple of weeks ago. And it said YY is deteriorating full stop."- interview 5

"she's even more brilliant because of her difficulties, I think they think she's even more special. If that makes sense"- Interview 10

"I think I was feeling more guilty because I had known before that there was something not right and I had let myself believe that everything was fine and I was more annoyed with myself for listening to you."- interview 9 mum

"I very much thought it was from me because of my epilepsy, so I assumed right from the start that that was where it had come from. So that bit would be the bit that had concerned me or had made me feel a little bit guilty" – Interview 7

"we are in a special little gang"- interview 6

"your fears are confirmed. Then you go through a process of like grief really. That's how I can best describe it. Then you think that the path your child is going to take." – Interview 4

"I had very bad post natal depression ... I had gone to see somebody and they gave me pills and I didn't want to take any pills, I was so resistant. Not only was I against the whole medication thing, I was worried about how it would affect my milk and so I fought it for absolutely ages and my degree is in psychology anyway so I wanted CBT and they said you'll be put on the list but meanwhile take these. Anyway, I was never offered CBT"- Interview 4

"went around in a bit of a mush of yes she has this gene but what did it actually mean?"- interview 1

- Living in a "bubble"

"to a certain degree probably life has bypassed because we are in this bubble."- Interview 8

"lived in our own little bubble but I think we've got a different attitude towards it now, we know what's coming, we know what to expect. I think the before we just stayed where we were and just got



on with it day to day, because it was a wee bit scary thinking about 10, 15 years down the line but now that you've seen kids 10, 15 years older than YY we're like well that's perfect, we know what's coming"- Interview 9 mum

"It only sunk in when we went to the conference really. WE didn't really know until we went to the conference what was... what was going to come for us in life and that that was the best thing and that was probably the point when it kind of sunk in for the pair of us. And it was like right well we know what we've got to do now"- interview 9 dad

"I think I was worried that I was just in a bubble and then it would hit me that I would see all these poor children and think oh my god, the future is bleak, but I don't know I was anxious, because I remember driving there and kind of every so often go oooh... and bursting into tears but I don't know why, I mean it was great going there, it was good, it was really reassuring"- interview 10

"If more medical people were aware of how to point families in the right direction, ... we were still a bit like -where do we go, where do we turn, who do we look to?... we kind of just lived in our own bubble"- interview 9 mum

"to a certain degree probably life has bypassed because we are in this bubble... I know that I've lost a lot of my confidence now and like when I do go out for a drink, I don't enjoy it"- interview 8

"in my bubble actually probably doesn't do us any good at the moment when we get to a wee bit bigger bubble then maybe we'll take some of that on board but at the moment that's not what we need for YY" – interview 5

- Coping mechanisms

"she's my baby and she's brilliant so it doesn't matter"- interview 10

"I was on anti-depressants ...I kind of bottled it up... Thinking back now I really genuinely think that had XX had no problems and been a normal baby then I don't think I would have, I can't see why I would have been. I think it was because of all the worry and concern about the condition and that what did bring me down."- interview 2

"you kind of regroup and reassess and realise they are still you're children, it's nothing they've done. They're still wonderful gorgeous people and they are all the more special for their difficulties and especially now they are getting a little bit older seeing how they face everyday life and going to school... Just seeing how they overcome everyday challenges that most children just take for granted, you know being able to talk or run around with their friends and everything else. It's a humbling experience; it teaches you a lot about yourself."- interview 4

"I would have kind of scoffed at that kind of thing before and thought oh some people get their kicks out of being part of these big charities and it's not, I tend to be more kind of keep myself to myself and my social group rather than kind of meeting complete strangers"- interview 10

"But I just try and think of YY as his own person. And just because he's got the same syndrome as these other children, doesn't mean that he is the same as them or that the same things are going to happen to him, so we just keep trying to think like that"- interview 6

"I understand that it's, I don't know if they class it as life limiting or well because the medical papers I've read. Of the 13 children that they did the study on, 10 of them had died before they got to the age of 5"- Interview 6

"because you have to cope and actually it's quite hard when you've got children because they don't understand that you're going through this process ...actually for their sake you just need to get on with it... It's like finding Nemo "just keep swimming, just keep swimming" and you do."- Interview 5

"I think when you get locked into thinking and focusing in on the things we need to do, that kind of keeps you going rather than worrying so much about"- Interview 2

"try not to discuss what's going on unless it's to the forefront. And it's on the calendar. It's just our way of dealing with it...I don't know whether I could cope with it at this time ... There's a whole batch of stuff that's going on at the minute. And probably that would just tip the scales for me."- Interview 1

"There was a whole mix of emotions. You still have to get on with life. You've got all these other bits going on in the background. It was difficult. And it's still difficult"- Interview 1

"we didn't speak we just didn't manage...juggling too many balls. You just get dragged down."- interview 1

"I coped ok. I'm quite kind of logical and scientific... I've got a close friend of mine has got a disabled daughter and she doesn't cope with it very well at all. She is constantly depressed by the fact that she has a disabled child. Whereas I don't know what it is. Maybe it's just my general positive outlook on life. I just think that I can't change it so why stress about it."- Interview 6

"Just take it a day at a time. You can't plan too far ahead because you don't know, even when you know what's wrong with them you know all the children are different... You just take each day as it comes and just enjoy them."- Interview 3

"she's even more brilliant because of her difficulties"- Interview 10

"I just kind of more accepting that this is what she has and she's my baby and she's brilliant so it doesn't matter"- interview 10

"but other people do it, and other people get on and if it's your child then you don't think about it, you've already done it for until they've been diagnosed and life is just going to carry on as normal for you.... there is always going to be one person that you'll think. Thank god it's not as bad as that, as bad as that sounds. "- Interview 9 mum

"we've made some good friends who've got children of all abilities and actually seeing their children who aren't as able as YY is actually quite a good test of actually life could be a lot ... He is loving, he is full of life, he enjoys life and he is happy at the moment. We're just trying to give him the best start we can. ... I think if it had been life threatening I think that would have been a whole different kettle of fish. I think that must be a whole different ball game. I couldn't begin. And I do have friends who have children who have life threatening diseases and disorders and I just, I don't know how they do it. So, that does help in that respect. You kind of bench mark sometimes, actually we are really lucky and YY will have a good life and he will have his challenges but you know hopefully he will be independent and happy as he will be and that helps." - interview 5

"everything might come crashing down but today he's my little boy and that's what life is for him so you just get on with it so yeah" - Interview 8

"maybe everything will come crashing down" - Interview 8

"I'm fairly laid back about it all. I've always worked on the fact that he is very happy and so I just think as long as I've got a happy child" - Interview 7

How they felt the healthcare team managed the family included:

- Hospital appointments and healthcare professionals

"We kind of stopped going to a lot of clinics that I didn't feel we were getting anything out of. I made the decision that we need to get off the wheel. ... And she needs to live a wee bit of life and not keep her in hospital." - Interview 1

"I know I don't feel like I turned away any help I think I took all the help I was offered... But I feel everything was offered because I kind of asked for it. Like the physio and the speech therapy" - Interview 2

"I discovered that there is a \*\* clinic in Yorkhill and I literally had to ask to be referred there. And Yorkhill had been great, in that you know the doctor, it transpires that the consultant when he met YY, he met YY on 2 occasions and he commented that he had never seen a child like YY and so now we are going through investigations again to see if there are other issues alongside his \*\* but he says he's never sort of seen a child like YY. He's definitely not following the norm even for Prader willi, so we are kind of in a sense going through it again" - Interview 8

"It would be really really beneficial to have one in Scotland, to have a sort of 1 stop place that you can go, so for instance if the diagnosis had been made then you get an appointment for this 1 stop clinic and in one place you would see the cardiologist, the haematologist and the speech therapist and whoever you need to see, do all that just in one day." - interview 4

"he basically gave us the diagnosis, he said go away and think about it and then we'll get you back in a few months time and if you have any questions and you know we can have further discussion then so that was fine we went away, we didn't, we hadn't even heard of it before so we couldn't have asked any questions that day...I don't know why it took so long to go back to genetics to actually

discuss it because now that since going back there a month ago, we obviously, we know everything about it, you know, we've already lived with it for a year so we knew everything about it by now so we actually had nothing to say to the genetics"- interview 11

"The paediatrician and the cardiologist tried to sort of amalgamate their appointments, yeah they tried to amalgamate their appointments so that I wasn't having so many appointments because I was just ending up with loads and loads but that didn't work because the paediatricians were always running late and the cardiologist always seemed to be on time"- interview 11

"I think they should have a professional, even if its a case of a 10 minute appointment with a liason officer or somebody who can just explain things a wee bit more. I think the time in between getting a diagnosis and actually getting information was too long and they didn't give us enough information, they didn't give us any information when we left that room that day."- interview 11

"if somebody is coming into their care that has a syndrome that your not familiar of, I would possibly like them to read up on it first, get a better idea, a better understanding of it"- interview 11

"I think everything was explained you know quite clearly at the time and I saw them on going so any questions that I had, which I may have had and I probably did, I would have just asked them in the course of seeing them routinely."- interview 10

"we would have at least 4 days of appointments sometimes 3-4 appointments in the 1 day. We were in ninewells at least twice a week just because we saw so many people. There was the physio, the speech and language, paediatricians, dieticians, eye appointments, ENT appointments... it would have been helpful if there was somebody t over see the appointments and try to plan it out a wee bit better"- interview 9 mum

"half the doctors had never heard of it that we were seeing and half the people that we seen were like looking it up before they came to"- interview 9 mum

"team around the child meetings. That's been helpful"- interview 9 dad

"There is no real dialogue or conversation and I think I know that a couple of doctors are slightly frustrated with me because of the amount of reading that I do about the syndrome and because of the amount of the research that I am aware of."- interview 8

"we are seen in Glasgow, I think that's just every 6 months just now that we go there. Yes I certainly wouldn't say he's not looked at often enough. I would say I'm quite happy with the way it's all monitored."- Interview7

"I'm a big supporter of the NHS. Being as lucky as I am"- interview 7

"I didn't know the difference between an orthopaedic consultant and an orthotist and a occupational therapist"- interview 5

"I suppose I'm kind of, not leading the care but asking them what they think about the developments + maybe not aware because there's maybe not that many children and they can't be reading all the research papers all the time can they, you know find out what's going on." – interview 3

- Meeting the parents' emotional needs

"we set up a team around the child meeting so we've had a couple of them and we've agreed to start doing that separately and things again and then meetings have been amazing absolutely brilliant just to have everybody involved, you know instead of repeating yourself and maybe missing something out or miss hearing somebody, everybody's there, you just speak your mind and somebody will answer you" - interview 11

"somebody who came into those types of meetings with me and then afterwards I could kind of almost bounce off of and kind of say "that bit there, did I hear that bit right" other than my husband, I think somebody who wasn't in a sense personally involved" - Interview 8

"WE had somebody from parent to parent but if I'm being completely blunt, it was quite a confusing. ... I think I kind of acknowledge that at the moment I'm, I probably seemed as if I was coping far too well with it because I was putting a front on it and I didn't want to admit to people that inside I was slightly heartbroken" - Interview 8

"we didn't get any leaflets of anything or anymore advice. It was just we had the genetics appointment... maybe it would have been good to be able to talk to somebody else about it after I had then gone and done a bit of research... I suppose it might have been good after I had got the diagnosis, to get a phone call from somebody a month later to say how are you coping with it? How are you finding everything now?" - interview 6

"Waiting for my test results and having to e-mail and phone was really not the best way because I didn't know when I was going to get this news and it was worrying me that was I going to be in the school playground when I got this phone call and what was I going to be doing if I had the kids in the car and silly little things like that make you worry" - interview 5

"didn't get a lot of empathy...whole weekend was just horrendous" - interview 2

"My concern for us is, yeah we were told all the bits, like in the letter and stuff but we've not been directed to any place like a group of people that have it." - interview 1

"go and seek counselling. From somebody that knows more about it. That could maybe put all the bits together that you want to know. And it would have been nice if there had been a group of people, or if somebody else had had it to say my son or daughter is now this age and it would be nice to kind of get a bit of feedback from other people. Folk to say look it's achievable. I'd like to know a bit more about where are we going." - interview 1

"Once you left the clinic you were on your own basically. And I just felt at that point I could have done with some kind of follow up to see how we were; apart from just clinically but emotionally." - interview 3

"seeing a different consultant. I was like I don't want to see you. I'm really sorry but I see the consultant that I was already seeing. Because I'm not going to sit and explain XX's whole life all over again. And that was the difficult part for me." - Interview 1

- The impact of the rarity of the condition on hospital management

"My doctors don't have the slightest Scooby" - interview 11

"I understand that students have to train and these are really interesting cases and things like that. But there is a couple of times where because I was so emotional about the whole thing and I didn't want anybody else sort of there, this was my baby and I didn't want people sort of standing looking on and just being very academic about it...there were a couple of times that I was asked when they were in the room and I just think that's not consent." - interview 10

"you wanted to be upset but you weren't quite sure what you were being upset about" - interview 9mum

"half the doctors had never heard of it that we were seeing and half the people that we seen were like looking it up before they came to" - interview 9 dad

"nobody's ever met another child with the same thing. So it's not like they can say "well in my experience of children with this syndrome we do this"" - interview 6

"go with your own instincts the doctors don't always know best." - interview 6

"Because shes rare people want to see her. And I'm kind of at the stage where I'm like no, I need this wheel" - interview 1

The support available for families included:

- Introduction into a "new world" and support required

"you go on there and actually you are in this other little world and Holland is a beautiful place but it's not Italy and there are times when you can feel a bit isolated" - interview 5

"you are just thrown into this world, it's like you go through a time warp" - interview 5

"through the post natal depression, the addenbrooks score. Each week she would give it to me and each week it was even worse and she'd say you need to go and see somebody and I'd say yeah you know what you're right but it's just your mum, you're bottom of the pile." - Interview 4

"Obviously if I could change it I would instantly but I can't so it's out of my control so you've just got to get on with it." - interview 6

"I think I very quickly then just went into like, I've been told since then that I'm a very professional mum" - Interview 8

"I was a project manager in my former life because I could just say right ok project and I just went into project mode and I just went right ok. What do we need to do does he need- this this this, right who does this. But a lot of other parents may not have that ability to do that. I was very lucky to have some of the skills to be able to get out there and to arrange some of this. Where was that hole? And who do I need to speak to and right I'm going to speak to them now and I wouldn't let it rest until we had everything and it took about a year-18 months to get things in place so that we felt as though YY had a good team around him and got the right support for him." - Interview 5

"there was just so much going on I just had to constantly remember that there was a baby in the middle of all this. And regardless of what the matter is. He still needs to be fed and loved and all that sort of stuff." - Interview 8

"very quickly you're kind of, you're just completely thrust into this world of what does it mean for the future and a lot of things obviously had to change for us." - Interview 8

"I started off with the attitude that we really didn't know if she'd walk very well or if she could be really quite delayed in walking and talking and if she'd even be able to go through main stream school but, she ticked all those boxes really quickly. I think it was a combination of just what she was able to do and encouragement we were able to give her... I think when you get locked into thinking and focusing in on the things we need to do, that kind of keeps you going rather than worrying so much about" - Interview 2

"we said "what do you think you want to write and what do you think about your body, what do you think is great?" and it was fantastic the first thing he wrote on the bottom was "I like myself" and I just thought brilliant" - Interview 5

"functional things like other parents were speaking about things to stimulate their fingers that they'd use for their children and just things like dinner forks and lengthening light switches and those kind of things to make life easier so that's handy. Just peoples different experiences and homemade gadgets and contraptions" - interview 10

"Only at gala day when there was little girls dressed up and highland dancing. She wouldn't be able to get her hands up to do it" – interview 10

"Obviously well I had to give up work" – interview 9 mum

"he's got a blue badge and then you go in a parking space and you get people looking at you like- you're not disabled – and you're kind of like actually my child is but he is in a buggy" – Interview 8

"if someone could give us a bit of information or sign post us then that may be something that we could look into" - interview 1

- Financial support and housing

the thing that for a long time upset me more than anything was the thought that it's going to be a struggle for her to lead an independent life ... I think it's more important to find a role in life, be it a job or working in some way that she'll get something out of life in that way. She needs support to do it well. And hopefully we'll be there to do it. And that's obviously another worry that, we'll be getting older and who'll be able to help her when we're not basically you know further down the line. I suppose that's more of a concern. ... we have started saving money away every month for the future, for whatever the reason we might need it for, we had money going away every month." - Interview 2

"its nice because theres a lot of sort of fundraising things that goes on for XX and everywhere that she s been, just because its such a small town its easier, everybody knows." - interview 11

"in Yorkhill they did introduce us to the family fund because we were taken down in an emergency situation we obviously didn't have time to save for it etc. And they awarded us no problem at all with some money to keep us going while we were down there." - interview 11

"parent to parent support worker so she comes in quite often and when she first came in she made sure that XX was getting all the benefits she was due. This year we've booked a holiday and shes actually asked for funding for us for the holiday because XX is entitled to it." - Interview 11

"we've got an average house. ... stairs up to a bathroom and the 3 bedrooms. And then it was well we've got son that can't walk... there is only so much they can do and we can't afford the rest and they would only give us 20% towards" interview 9 mum

"You almost think we don't need it now but we know we are going to so should we start fighting now or do we wait until we need it and then start fighting and then are we going to have more of a fight on our hands." - interview 9 mum

"My husband has basically had to go self employed so he can get time off for appointments without fear of losing his ... we're kind of stuck in limbo with our mortgage, we're in a kind of 2 bedroom house and we could ideally do with a three bedroom house ... I've had a mortgage since I was 21 years old and I've worked really hard for it and it is quite frustrating and upsetting to think that I could lose all that." - Interview 8

"You just feel like you're constantly justifying that your child is disabled" - interview 8

"it didn't make any difference financially me working, I was actually better off on benefits, so I just thought I'll just see how it goes" - interview 6



"I feel that I'm lucky. My husband can earn enough money that I don't have to work. It must be very hard to have that balance between being able to give your children what you want versus having to get money to be able to live. But in terms of, silly things like petrol and the number of trips up to hospitals and having to buy equipment because there is no money and stuff." - interview 5

"I didn't feel like there would be a financial problem ... are you not getting disability living allowance? I thought I won't get that, she said don't be daft you've got a named disorder, she will get. I didn't know anything about that; that was something I needed to be told about. It was complete news to me. + I remember thinking at no point had anybody in the hospital"- interview 1

- Information

"Even though it meant, it was going to bring a lot more problems with it but, rather knowing than not knowing yeah." - parent 2

"we left well I left that appointment with very little information. So it was, you wanted to be upset but you weren't quite sure what you were being upset about. If that makes sense, there was something wrong but we still had to go and find out.... It was just a name floating" - Interview 9 mum

"foundation was the main place we were directed to and it is quite good foundation and they send out a lot of information so it is a good thing to have" - interview 11

"being given the diagnosis but not being given any information there and then to take away with us, you then automatically look on the internet and on the internet you automatically find the worst case scenario everytime, ...If I had been given 3 leaflets to read I wouldn't even have switched on my computer"- interview 11

"I don't know if you ever saw that \*\* documentary...I watched on catch-up over the internet when I was pregnant after they'd kind of said, that was my friend that had told me, and that's she's just an inspiration that wee girl"- Interview 10

"well some of the stuff is complete gobbledigook especially some of the genetics stuff"- interview 5

"I came out in floods of tears and we were given a sheet of a4 paper"- Interview 9 mum

"I do read up a lot or research"- Interview 8

"have looked into it, but I haven't studied greatly because I don't think there is any point in worrying." - Interview 7

"I've done lots of research, read lots of medical papers and been in contact with lots of other families with children with the syndrome." - Interview 6

"looked everything up on the internet and it was all quite pessimistic and not very good news... there is less information out there because it is so rare"- interview 6

"we were given this whole load of stuff. Paper they'd obviously printed off the internet. But it had come off the American site ...worst case scenarios ...horrific stories of people with horrific problems they've experienced."- interview 2

"initial diagnosis when I was pregnant, I didn't feel we got a lot of information then I have to say. I can remember I was kind of left to deal with it."- interview 3

"I actually probably wouldn't have wanted as much information as he had given me. It made me demented ... Those couple of months until she was born, because the information he gave me was you know really quite horrendous...he gave me print outs ... That was I think diabolical" – interview 3

"I mean I joined the association within a few months of the diagnosis. They were quite helpful over the phone... magazines and their websites...all the information I've had I think has come from there....it's good information, information for individual parents, all sorts of leaflets and packs and separate bits of information to give to schools and carers + teaching techniques + the information I've been able to give to them" – interview 2

- Using the Internet

"Yes I have looked on the internet. But to start with we were advised not to look on the internet. Because, I understand why because some of the sites can be more upsetting"- Interview 7

"I've been in touch with more of that side of the families with that syndrome and that's all via facebook which is amazing. And there is about 16 of us I think in our little group. All with kids with the same thing." – interview 6

"there's 2 support groups on facebook that you know its like, they just post something up and your like yeah you know I kind of relate to that and you can look and see what other people have commented on"- interview 11

"I would say that the facebook support groups been a massive help."- interview 11

"its real people and everyday life and its like what you'd maybe comment on facebook you know... I would direct anybody there before I directed them to the foundations page."- interview 11

"the internet is wonderful and not s wonderful because you do look up things and we did and it was that, that was awful."- interview 10

"look on the internet so everything I think that we know, we found out ourselves ...we left well I left that appointment with very little information."- interview 9 mum

"we could find were all from America and you know what they are like, they blow it out of proportion and have all there sad songs in the background and so we never really got an understanding of it"- interview 9 mum

"Facebook is amazing. Just it's like this wee family... it's always somebody who is going through the same thing or has been through it or is interested"- interview 9 mum

"we are actually on facebook and we do speak but we rarely speak about the kids, it's just every now and again how are you doing?... I'm kind of quite happy with the being on the forum if I've got any questions and these people can give you advice and I'm quite happy with that rather than seeing"- Interview 8

"one can drive themselves loopy reading all this and could he have that? And has he got this"- interview 5

"We tend not to go on the internet. ...I was like write it in a letter."- interview 1

"I looked at it on the net and kind of made myself unwell with it"- interview 1

"right away, we went home that day having spoken to the geneticist and we looked it up online and there was a picture of a child and that was her."- interview 3

- Where the parents found support

"for being such a rare condition it was strange. There is a lady that I know in the village who when she was diagnosed and I mentioned it and she said "Oh my next door neighbours brothers daughter has that and they have diagnosed sort of a year before". ... because I was still pregnant at the time and how do they cope and things like that, so that was helpful as well."- Interview 10

"that support group as well, you have to prove that your child's got Williams syndrome before they let you join like you have to send a photograph and explain how she was diagnosed and things like that so that your able to join the support group, so that, I can see in sense why they do it, so that some of the information can be quite intense and if you were only kind of guessing that your child had Williams syndrome and were on the page you would maybe follow steps that maybe shouldn't be followed. So I can see where they are coming from. But to somebody whos just had it diagnosed and think right I'll go there and have a look, it's a lot to maybe to do... they send you a lot of good information and they even sent me you the little red books babies get once their born, they sent me a little blue one"- interview 11

"it's a charity thing they run, they had a, it just so happened that they had an event up in Scotland when she was about 6 weeks old. And well I'm not really into this kind of thing but we'll go along we'll see what it's like and it was so helpful. There was tonnes of little kids there right through to teenagers that had the condition and they were just functioning like ordinary, anybody else but just did things differently and that was really helpful I guess."- interview 10

"the one thing that would tell anybody who's just had a diagnosis is go to the conference. It's the best thing you can do. No matter how scared you are ... you've got to almost face it some time"- interview 9 mum

"I have been given numbers of various people I could contact or various websites to go onto but I've never done it because I don't think it's particularly serious at the moment."- Interview 7

"So there is that kind of parent support group. It's unofficial but it really helps"- interview 5

"it could have been my own searching on the internet I found a support group"- interview 4

"you needed a more consolidated approach to it. I don't know. Rather than it just being sort of picked up by luck because you've got something else going on. Maybe there needs to be a sort of, almost a sort of checklist to go through. Have you been in touch with the relevant support organisation and have you been offered counselling. Even if it can't, even if there aren't funds available for it to come through from the NHS, even a sort of self help check list, so this is where you can go to get some help."- interview 4

" We have a conference with a couple of nights in a hotel. You've got the networking as well, and the informal discussion which is probably what we go for rather than the lectures ... I mean that's good too but I think it's because we're up in Scotland and there's not many children up here have it, and certainly nobody that I'm in touch with that I know of in the local area... meet up with other children and see how they're getting on Compare notes with the parents you know how their coping with the transition to high school. " interview 3

- Education and Schooling

"I've spoke to the local playgroup that YYs at and they are actually quite happy to take her and they will put the extra support in for her"- interview 11

"because of the experience we had with the private nursery ... kind of just then I sort of lost my faith almost in private nurseries and very much. So I know that he's going to get the support he needs"- Interview 8

"he is incredibly bright. His speech is, he keeps up amazingly with his speech for the age"- Interview 7

"He went to the nursery of this special school ... It wasn't a big drama really, it was just he was going full days instead of half days. He was going to the same place. He was familiar with all the people there. It was good. He was enjoying it."- interview 6

"actually we wanted him in a very small traditional village school and he has blossomed there and everyone just accepts"- interview 5

"when it comes to education. For my boys, particularly the older one it's kind of so specialised. It's as though, I mean I haven't met anybody yet who can, who has the same sort of picture as how he has

presented which seems to be real speech difficulties but maybe his IQ is down a bit but he seems to be holding his own at school and that side of things. But we looked at a school for him, thinking, the head teacher of his primary school said to us when he gets to P4 which is what he is in now, you will notice a real shift in the teaching and I'm voicing my concerns now that he won't be able to cope, we won't be able to meet his needs in this setting in this school. So we went off to look at a school for deaf children which I think half of its pupils were deaf and half had severe communication difficulties. However, in the place of a healthy boy, most of the communication difficulties are in the autistic spectrum and he clearly is not part of that, he is very very sociable, in fact his friends are probably the most important people in his life. A situation like that would not be appropriate because he is neither deaf nor autistic so he would find it very difficult to find a peer group in a setting like that. So, we have kept him where he is, in the absence of knowing anything better and we'll just have to see how this year goes and how he copes." - interview 4

"it would have been hard to give the information I was given to help the school on the just general education side of things. If there wasn't an association there or there wasn't help out there. I know the hospital are more geared on the physical and the dietary side of things than your emotional side of things and the educational stuff so, it's just getting from both sides really the help" – interview 2

"we're not quite sure how 2ndry School will go for her, because they don't thrive so well at 2ndry but as to what help she'll get then. Down the line if I need to change my hours to fit in with what she's doing, I don't know...at school she'll pick the healthy option, she won't pick, not that she gets offered it anyway, but any of the fried stuff...they've always known the situation" – interview 2

"The school's been great. They decided to give her a calculator in Primary 7... I've given them the leaflets ...only trouble we have at school is the low muscle tone. It's getting that through to them. It's not that she doesn't want to participate fully in all the physical activities. It's that she can't because she gets so tired so easily and she's not got the stamina...We had a letter from, we had a letter sent to them by the physio which helped and the school doctor had a transitional meeting to go into high school. The school doctor was really good and she made it quite clear that this is what all the problems were before she went up." - interview 3

- Respite care

"I can't leave her with a childminder and to be honest, not many child minders would be willing to take her on because she is a lot of work ... I don't know where I'd be without my mum." - interview 11

"my mum looks after him but she is now 70 and she is actually in hospital today having an operation, so she is not going to be able to look after him for the next while but he goes to respite... I can go off and do what a normal 30 year old would be doing" - interview 6

"luckily found a child minder ... SO she was able to deal with disabled kids." - interview 6

"not knowing who was there to help to help me to find the right place for him." - interview 5

"we've never had a problem with joining the clubs before but this time, we were going with a certain holiday company who pride themselves on having a very good child care facilities ... they refused to take ZZ, the 6 year old into club and they knew that, well they are a British company and they are employing British nannies and they should come under or comply with, it's not the disability discrimination act, the equality act, however because the service is being delivered in Portugal they didn't have to comply. ... He goes to a main stream school .... "oh, but if we've got one nanny and 15 children in club they won't be able to give him that attention"." – interview 4

"It's difficult to get a babysitter for a child like that, so that's still kind of held me back + child care has always been an issue. We had to hire a nanny when she was younger, so that I could go to work" - interview 3

- **Family and friends as support**

"we'd be lost without your mum" - interview 9 dad

"I don't know where I'd be without my mum." - interview 11

"My sister is a medical writer so you know; she kind of helped and explained medical terms to me." - interview 6

"the network of parents with additional need kids comes into it because you can say things to them that I couldn't say to my mother and that's the thing I don't really talk to my mum about how I'm feeling, sometimes she catches me on a bad day." - interview 5

"they were there when I came home and it was just a bit surreal actually, it was a bit surreal, and they were really good because they just took the children so they just left me a bit of time to breath and compose." - interview 5

"I suppose I don't even remember speaking to my husband about it ...just didn't discuss it in that way that it was making me feel that way ... I thought I don't want to add more pressure on to him that I'm not up, I'm feeling bad about it or not coping with it. But I probably did it to myself to be honest because I didn't talk about it" - interview 2

"we just knew that we've got a family network that would support us. We had the support there, so we are quite lucky there" - interview 2

"I had to refer myself to social work which took 3 years to get an assessment...really struggling with work my husband and her and I needed some respite... they need to get social work services involved right at the start. Somebody needs to refer these children to social services right at the start, you know so that when you realise you've got a problem, you've got somebody you can turn to." - interview 3

### How a genetic diagnosis affects the family included:

- The impact of conditions being genetic

it was just so hard to come to terms with the fact she might have it as well ...so it was really important that I got tested to see whether or not I had it and whether or not (my daughter) could have it. ...it's quite funny you feel really guilty that I really don't want (my daughter) to have it but (My son) has it and it's really, you just feel that you're not wanting to take or detract from the child that has the disability, you know have the disorder but I really didn't want (my daughter) to have it." - Interview 5

"we separated when I was pregnant but he had a slightly strange gait and walked on his toes a bit ... we did have a discussion with the neurologist ..And I think it was said no no it won't it can't be because there's just not the family history there because he was saying no no everything was fine everything was fine." - interview 5

"she was worried that, my partners a twin, their dad's a twin so she was worried that the other twin would possibly carry it" - interview 11

When we got the diagnosis for our immediate family, we immediately told my mother in law because my husband's brother, her other son, so she just has two boys, now he has various difficulties ... I don't know maybe she was just in denial or what but she was very resistant to what we were suggesting. And since then, however many years we are now on, she's been back in contact... when I last mentioned it, it wasn't sort of oh you know, I wasn't trying to be difficult or anything, I just thought it might give someone some sort of answer or certainly a lead to investigate further anyway and the impression I got was it didn't come from our side of the family... it was a dirty secret. It's nothing to be ashamed about why and try and sort of push it under the carpet, be open about it" - interview 4

"husband looked at the information and he thought we should have done, which did lead to a lot of problems and we are now divorced... he couldn't cope with my daughter and that she had this illness that possibly one of us gave her but we now know we didn't pass it on...But he wasn't really coping with her and her in and out of hospital...sent him quite far back and he got quite depressed and ended up he was suicidal" - interview 3

"My sister was pregnant when XX was got her diagnosis. And she was obviously it was genetically linked like through our mother. She thought it might be linked to her." - interview 1

"mum was definitely in denial for a good few years...she'd say oh she'll grow out of it sort of attitude... I tried to explain so many times" - interview 2

"they were in denial for most of the time really and they still really don't think that there's a lot wrong with her that some good, better parenting I hate to say would put right" - interview 3

- Family relationships

"I kind of bottled it up, so we're not kind of the sort of couple that'll sit and pull it to bits and think about it. We kind of did the opposite; we went opposite direction with it. ... I think it's just one of those things that we accepted it fairly quickly although you still have trouble dealing with it we both accepted it." - Interview 2

"then they'd go "oh what's that" so then you feel like you have to go way back and explain everything... if we said he'd been diagnosed with Down syndrome, then people they know what it is." - mum interview 9

"grannys and granddads probably actually took it a bit worse than we did to be honest" - interview 11

"wonderful, well he doesn't really know that there is anything up. I mean he hears us speaking about it and things like that and saying oh be careful when he's grabbing her. But he just, he just thinks she's amazing. She brought him a Buzz light-year when she was born so she was in there" - interview 10

"my mum, she was away in Paris recently looking at some church and praying for her arms and things like that" - interview 10

"my dad he took it quite bad to begin with. I think he's, I think he still can't t this day still can't quite believe it that you know it's not so much happened to me but it's that's his grandson" - interview 9 dad

"I don't feel I've got the energy or the time to kind of be counselling them to a certain degree." - interview 8

"As a grandparent you want to spoil your grand child by giving them chocolate and by you your child going up and hugs and reciprocating and that, YY can't do that. When YY hugs he bites and he pulls your hair, so it's not a hug." - interview 8

"My mum and dad, I told them specifically you must not Google, I will find the right information because they would be, especially my dad they'd be in there ... so I kind of drip fed them the bits that I thought was relevant for them to see" - interview 5

"His sister got very, she was getting up a couple of times a night and just coming into our bedroom and just wanted to be put back to bed. And she just went through a phase of doing that. ... So she is picking up about things. So you do have to find ways. Either you can find a bit of time to yourselves to allow the grieving process. But that's not easy; you need to have that kind of smiley faced mum at times." - interview 5

"pre-implantation genetic diagnosis, that brings up its own moral implications on its own. We weren't just focused on completing our family but also the boys were so much younger and we didn't know how they were going to grow up and how they were going to be affected and there was also that sort of, I hesitate to use the word burden, but later on in life as my husband and I get older, it could be that our eldest would have 2 siblings to be responsible for and care for." - interview 4



"Went through a bit of bickering... marriage counselling. We were falling apart at that point." – interview 1

"he's been involved in the young carers and they've gone over the condition with him and they've done a folder and looked into the ins and outs of it, so he's had quite a bit of support over the years from them. I managed to access that." - interview 3

"we just kind of knew it was something we had to get on with together" – interview 2

"there was a couple of ones in the neighbourhood, who were a bit cheeky about her to him but they weren't. But xx wasn't at the school so he wasn't getting hassle at school about her" - interview 3

- Friendships

"I would rather be open and honest about it rather than having people whispering "Oh, why isn't he talking yet" and all the rest of it." - interview 4

"we can basically say its kind of like downs syndrome" - Interview 11

"I've got a small group of very close friends ... we've never ever had one negative thing said about her which is great." - Interview 11

"But I think friends just they kind of drifted away even before he was diagnosed... people who did find out it was more pity, don't like that... but then they'd go "oh what's that" so then you feel like you have to go way back and explain everything, you couldn't really go yeah YY has got angelmans syndrome and people would be like "right oh that's a shame" " - interview 9 mum

"I had a couple of friends who I believe were just inconsolable when YY was in hospital but they came to visit him once and never seen him again." - Interview 8

"The truth is, as much as I've made some really really great friends, in another life I wish I'd never knew them. But this is the life that we've got so in a sense I probably have lost a few friends that you know drifted apart and we are in completely different places but I've made other friends and there is a different type of understanding there." - Interview 8

"some people would ask you, some people would just ask you what type of cancer he had and you know, you're kind of like he's not got cancer" - Interview 8

"majority of them are friends so and they know why he is not walking and, so no. I wouldn't say, I certainly don't find it particularly awkward. No and because I think people are more interested in the fact that he laughs a lot and than the fact that he bum shuffles everywhere so no. I don't think I've encountered any problems yet." - Interview 7

"I've got lots of friends with disabled children and they kind of go, "oh why has it happened to us" and feel really sorry for themselves" - interview 6

"my family have been great really supportive... But I was married to YYs dad and he didn't cope with it at all and that lead to our marriage breaking down... So it did have a massive effect on our family. But my mum and my dad and my brother and sisters are all brilliant with him"- interview 6

"I do have a good friend who has question marks, they can't work out, he doesn't have a diagnosis and they believe it's genetic so she is going to need another set of new genetic testing so it's quite good. We can support each other having been through bits and pieces."- interview 5

- Family planning

"I thought this is what life gives you. It's a bit of a pain but you just deal with it as it comes. So the point where I was offered the abortion, I just thought no this is what we have to do and we are just going to see it through and you just take what comes. I haven't really thought about all the ramifications of it I suppose. But I didn't want to."- Interview 3

"We are not having anymore children so thats not a concern,"- interview 11

"I wouldn't want to have another child with additional needs and then not have I suppose the same attention to give to her and to give to my little boy"- interview 10

"we thought well if we're having anymore kids then we have to do it now before he's too big for me to be 9 months pregnant and carry him... I was always we will have it tested, we will have it tested and then it kind of came to it and thought there was no point"- interview 9 mum

"well because the tests showed that it hadn't or they were thinking that it hadn't come from either of us. That wasn't really a problem but one child is fine anyway so I'm not thinking about that."- Interview 7

"I think they've said to me it's 1 in 100. ... It wouldn't put me off having more, definitely not and even if you could, because they have just identified the gene that causes YYs syndrome, they could test while I was pregnant but I don't know if that would even make any difference because once I was already pregnant, if they then told me that my child had the same I think I would still, there is no way that I could get rid of it."- interview 6

"we had always wanted 4 and what would be our options t have another child and so he referred us to, well initially it was going to be London but he was able to get us into Glasgow and had PGD. So yep yes you that's how you came about, she is my little miracle."- interview 4

"situation where he wants to start his own family. He can refer back and say this is what mum and dad did all those years ago and I'm sure medical science will have moved on so much more... it can be you know cut out in a generation and you don't need to, I hesitate to use the word suffer but, if they want to have their own family then here is an option for them"- interview 4

"I was so focused on trying to have a healthy baby that it only just hit me much later on "did we really do that? Should we not have given that more thought?" Just as well we didn't at the time. I suppose as the boys get older and you see their characters grow more, you know just how special

and wonderful they are, and you kind of god you know, there were potentially 7 others, and you know it's just one of those things." - interview 4

"I was 28 weeks pregnant and I was offered an abortion which I was against immediately. Obviously because I thought it was far too far advanced to consider that no matter what the outcomes going to be" - interview 3

"We would have liked one more, but we've decided that theres no going to be any more because next time it might be getting more issues...That there is something else that may appear." - interview 1

"Daughter not given testing till 14 when she may already be sexually active" – interview 1

"and I know the chances of it reoccurring are like 1000s and the risk of it happening again were pretty remote... we did wait longer...we kind of thought is there going to be a problem with another one" - interview 2

"for her it's now 50 50 + we were given advice... I'd like to think that if we'd had her first we'd still have gone on and had my son because we did really want 3" - interview 3